GENOMIC MEDICINE IN PRIMARY CARE: TEXAS PHYSICIANS' ADOPTION OF AN INNOVATION

A Dissertation

by

SANDRA GAYLE SUTHER

Submitted to the Office of Graduate Studies of
Texas A&M University
in partial fulfillment of the requirements for the degree of

DOCTOR OF PHILOSOPHY

December 2003

Major Subject: Health Education

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Approved as to style and content by:

Patricia Goodson (Chair of Committee)	B.E. Pruitt (Member)
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ABSTRACT

Genomic Medicine in Primary Care: Texas Physicians' Adoption of an Innovation. (December 2003)

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New applications of genomic medicine stemming from the Human Genome Project are predicted to become routine components of primary care. Primary care physicians (PCPs) will increasingly become responsible for screening patients for inherited diseases, recommending genetic testing, and making referrals to genetic services. Clinical applications of genomic medicine will occur at a variable pace. Characteristics of an innovation such as genomic medicine are strong indicators of its potential for adoption. The purpose of this study is to assess whether (and to what extent) physicians' perceptions of genomic medicine as an innovation influence their likelihood of adopting this innovation into primary care.

The study's sample consists of 400 primary care physicians in Texas and employs a survey design. Based on Rogers' Diffusion of Innovations Theory, the perceived characteristics of genomic medicine – Relative Advantage, Compatibility, Complexity, Trialability, and Observability – are the study's independent/predictor variables. Likelihood of PCPs Adopting Genomic Medicine is the dependent variable. The nature

of the social system (private or group practice) is examined as a possible moderator variable.

The study suggests that Texas PCPs who are likely to adopt genomic medicine strongly perceive its clinical uses (such as genetic testing for carrier status or susceptibility to common diseases, testing an embryo for genetic disorders before it is implanted, and supplementing a family history) to be highly advantageous. For half of the PCPs, genetic services such as genetic counseling and genetic testing are not compatible with current practice. Perceived complexity of the innovation is the strongest predictor of likelihood of PCPs adopting genomic medicine. Many PCPs find it difficult to stay updated on genomic medicine and locate genetic services. Although Texas PCPs feel genomic medicine can be gradually incorporated into primary care practice, most are not presently observing their colleagues adopting genomic medicine or assisting their patients to make decisions regarding genetic services.

Future efforts to advance the use of genomic medicine in primary care will require more emphasis on genetics in medical school curriculum and continuing education programs. Links with specialists trained in genetic counseling and health education will be essential to translate relevant information to patients and families.

This dissertation is dedicated to my husband George.
I could not have done it without you.

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Compiling a dissertation can seem overwhelming at times for anyone. My experience was no different. My only fear now is to fail to properly acknowledge and thank the individuals who were instrumental in making it happen.

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I deeply appreciate the special efforts and expertise of Chuck Chiles for converting a huge electronic physician database into one more suited to the needs of my study. I am also indebted to Miki Collins for entering and organizing the statistical data for this study. Special mention goes to Norman Collins, Angela Suther, and Chris Suther for the hours they spent stamping, labeling, and stuffing envelopes.

Words are insufficient to thank my husband, George, and my children, Natalie and Christopher, for their boundless love and encouragement and for enduring this long process with me.

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CHAPTER I

INTRODUCTION

Knowledge, resources, and technologies stemming from the Human Genome Project have advanced understanding of the potential contributions of genes to human health. With developments and new applications of such knowledge and technologies to the medical industry, the field of genomic medicine has been born.[1]

Genomic medicine will increasingly be used to address common conditions known to have significant genetic components. Early intervention might be able to prevent some diseases such as hypertension, obesity, diabetes and cancer, while pre-conception counseling can be appropriate for diseases that are lethal, severely disabling, or untreatable.[2] Knowledge of the risk of a particular disorder will be useful for people who need to avoid the environmental triggers that convert gene susceptibility into disease. Another prospect of genomics, the field of pharmacogenomics, has the potential to predict which medications will be most effective for specific patients.[3]

Some health professionals dispute the claims that genomic medicine will revolutionize clinical practice and public health.[4] For these professionals, medical genetics is still considered the province of specialists who spend their time evaluating unusual cases of Mendelian disorders, birth defect syndromes, or chromosomal anomalies. However, according to Francis S. Collins, M.D., Ph.D., director of the

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This dissertation follows the format of *Genetics in Medicine.*

National Human Genome Research Institute (NHGRI), "That is all about to change."[5(p540)] The next decade will see genetics spreading rapidly beyond the confines of specialist centers into primary care.[3] While many primary care providers already incorporate genetic screening into their routine services, the demands on family physicians will increase substantially as new genetic tests and treatments become a routine component of medical care and prevention.[6] Within this scenario, "medicine will no longer be for the sick."[7(p123)] Comprehensive genomics-based health care will be standard preventive medicine.

Becoming familiar and staying updated with an ever-increasing number of genetic technologies will be a fundamental challenge for primary care because, as Williams [8] points out, most primary care physicians (PCPs) have only received an introductory genetics course in medical school. Most may not have the experience, training, or time to adequately order and interpret the results of complex genetic tests.

Adoption of a medical innovation (new ideas or technology) often involves perceptions that a new intervention will benefit a patient or a population.[9] For many primary care physicians, medical progress through new genetic technology has stimulated deep-seated anxieties concerning the sanctity of life. Potential threats to the rights of the embryos, fetuses, women and the disabled have become just a few of the major concerns.[10]

Primary care physicians are predicted to become genetics gatekeepers, "opening the gate" for patients by screening them for inherited diseases, recommending genetic testing, and making referrals to genetic service providers. However, clinical applications

of genomic medicine will occur at a variable pace for different disorders and in different areas of primary care.

Purpose

The purpose of this study is to assess whether (and to what extent) physicians' perceptions of genomic medicine as an innovation influence their likelihood of adopting this innovation into primary care. The likelihood of adopting genomic medicine into primary care is this study's dependent variable. Physicians' perceptions of specific attributes of genomic medicine are the predictor/independent variables. As both likelihood of adopting genomic medicine and perceptions of genomic medicine as an innovation can vary according to the social/professional system physicians are engaged in, characteristics of their practice (i.e., whether they are involved in private or group practice) will be examined as having a potential moderating effect upon the dependent and predictor variables.

Theoretical Framework

This study is based on Everett M. Roger's[11] Diffusion of Innovations Theory. The theory defines innovation as an idea, practice, or object that is perceived as new by members of a social system. The members of a social system may be individuals, informal groups, or organizations. The innovation in this study is genomic medicine and the social system is the formal structure (private or group practice) within which the physicians practice. According to Rogers,[11] the composition of a system will influence the behavior of the members of the system. Rogers defines diffusion as "the process by

which an innovation is communicated through certain channels over time among the members of a social system"[11(p5)] For the purpose of this study, diffusion refers to the adoption of genomic medicine-related services into primary care practice.

According to Rogers,[11] there are five characteristics of an innovation that influence the rate of its adoption. These are the relative advantage of the innovation, its compatibility, complexity, trialability, and observability. Primary Care Physicians' perceptions of these characteristics of genomic medicine will be measured in this study (see conceptual and operational definitions for each of these in the section describing the study's variables, below). The nature of the social system will be examined as a moderator variable, as it relates to the likelihood of primary care physicians adopting genomic medicine into their practice.

Variables

Based on the Diffusion of Innovations Theory, the proposed model in Figure 1 represents the relationship between primary care physicians' perceived characteristics of genomic medicine and the likelihood of adopting this innovation. Relative advantage, compatibility, complexity, trialability, and observability are the model's predictor variables. Likelihood of PCPs adopting genomic medicine is the dependent variable. The nature of the social system is shown in the model as a moderator variable.

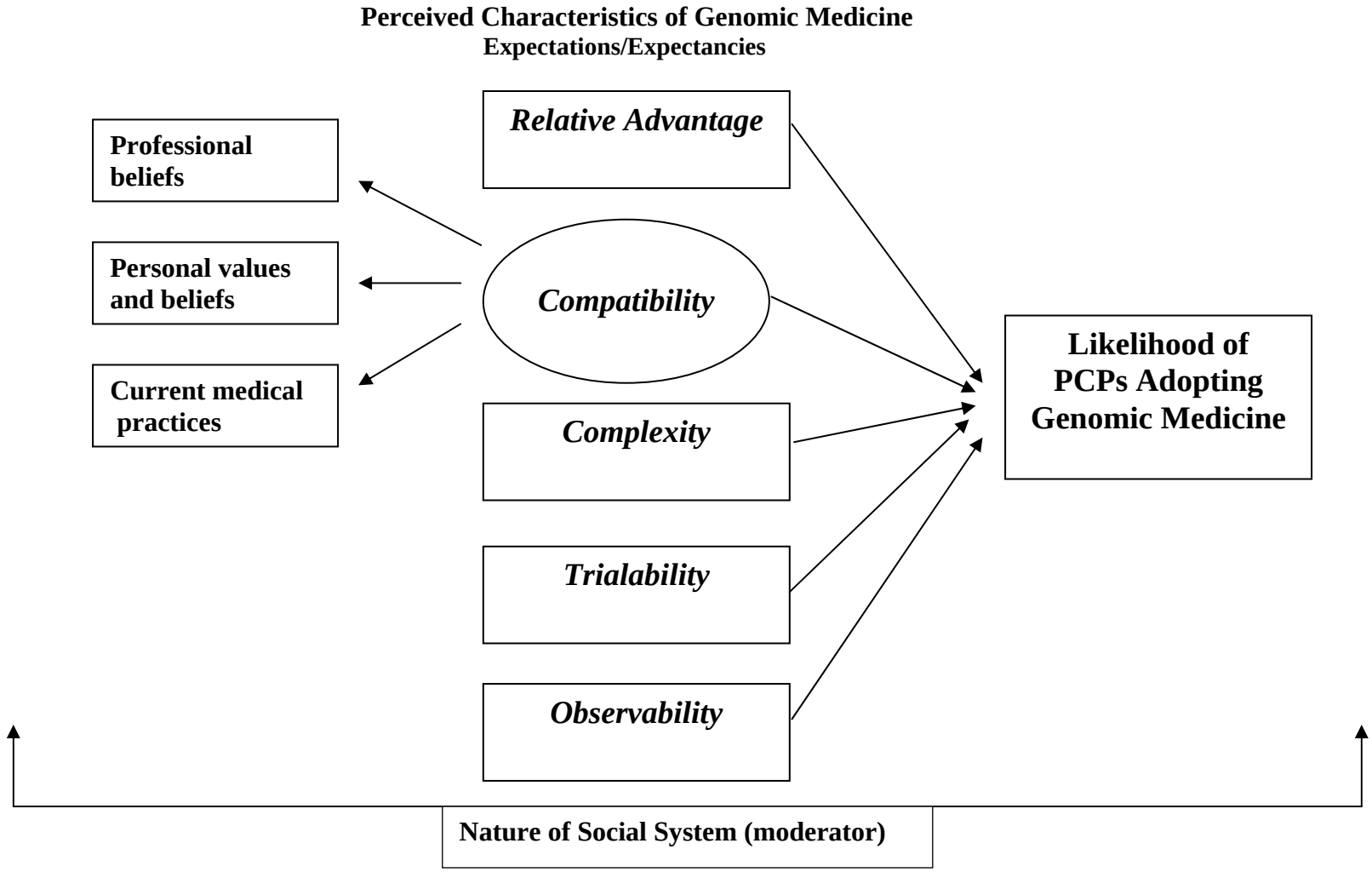


Figure 1 Model to Assess Likelihood of PCPs Adopting Genomic Medicine

Dependent Variable

The dependent variable that will be measured in this study is, "Likelihood of Primary Care Physicians Adopting Genomic Medicine." Conceptually this variable is defined as the likelihood that primary care physicians will incorporate new developments in genetic technology into their practice. Genetic technology includes genetic testing, genetic therapies, genetic consultations, and pre-conception counseling.

The dependent variable will be continuous. It will be measured by adding the respondents' scores from five items that question primary care physicians' likelihood of 1) ordering a carrier test for an autosomal recessive disorder, 2) ordering a preimplantation diagnosis on an embryo to check for genetic disease, 3) ordering a predictive test for a patient to check for risk of disease, 4) providing pre-conception counseling, and 5) referring a patient for a genetic consultation. The items used a 5 point likely-not likely Likert scale, for response. A lower score indicates a higher likelihood of adopting the innovation of genomic medicine.

Independent/Predictor Variables

For the purpose of this study, the construct "perceptions" is being understood as "attitudes." Operationally, attitudes are usually measured through two dimensions: beliefs (outcome expectations) and values (outcome expectancies).[12, 13] Expectation and expectancy questions were asked for each of the characteristics of the innovation of genomic medicine: relative advantage, compatibility, complexity, trialability, and observability. Multiplying each expectation and corresponding expectancy scores and

summing these products will compute respondents' "perception" scores for each of the characteristics.

"*Relative advantage*" is an important characteristic of the innovation that can influence its adoption. The degree to which an innovation is perceived as better than the idea it supersedes is considered a relative advantage.[11] If a physician perceives components of genomic medicine to have an advantage over what is being presently practiced, adoption will be more likely. For example, he/she may believe one of the advantages of genomic medicine is, "to diagnose a genetic condition in an embryo before it is implanted instead of waiting and doing an ultrasound later in the pregnancy." In this study, relative advantage is measured with 4 expectation items, using a 5 point agree-disagree Likert scale and 4 expectancy items (How important is it to you that…?), using a 5 point important-not important Likert scale, for response. A lower score for this scale indicates a higher perception of the relative advantage of genomic medicine.

"*Compatibility*" indicates the degree to which the innovation is consistent with existing values, beliefs, and present practices of potential adopters. Compatibility, in this study, is being proposed as a latent variable measured by 3 indicators: Compatibility of genomic medicine with primary care physicians' professional beliefs, compatibility with current medical practices, and with their personal values and beliefs. The more genomic medicine can integrate and coexist with primary care physicians' present technology and with their professional and personal beliefs, the greater its prospects for adoption and diffusion. An example of a compatibility item in this study for the variable compatible with "professional beliefs" is, "Termination of pregnancy

when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my professional standards."

The indicators "professional beliefs" and "personal values and beliefs" are each measured with 2 items for expectations, using a 5 point agree-disagree Likert scale and 1 item for expectancy, using a 5 point important-not important Likert scale. A lower score on these two scales indicate a higher perception of the compatibility of genomic medicine with the professional beliefs and the personal beliefs and values of the primary care physician. The compatibility with "current medical practices" indicator is measured with 3 expectation items, using a 5 point agree-disagree Likert scale and 1 expectancy item, using a 5 point important-not important Likert scale, for response. A lower score for this scale indicates a higher perception of the compatibility of genomic medicine with the primary care physicians' current medical practice.

A third construct measured in this study is primary care physicians' (PCPs) perception of genomic medicine's "*complexity*" or the extent to which the innovation is perceived as difficult to understand and use. Genomic medicine will most likely require additional training in order for many PCPs to incorporate this innovation into practice. Available genetic resources will also be a necessity. An example of a question to assess perceived complexity in this study is, "How easy or difficult is it for you to locate available genetic services?" A perception of complexity is measured with 2 items for expectations and 2 items for expectancies, using a 5 point agree-disagree and easy-difficult Likert scale, for response. A higher score for this scale signifies a perception of genomic medicine as highly complex.

"*Trialability"* is a characteristic of an innovation that refers to adopters' ability to try it out on a limited basis without total commitment and with minimal investment. Trying out or gradually incorporating genomic medicine into primary care practice allows potential adopters to reduce uncertainty about its risks and benefits. Many primary care physicians may feel genetic technologies cannot be incorporated on a trial basis. In this study respondents were asked, "How important is it for you to incorporate technologies that you have tried first?" A perception of trialability is measured with 2 items for expectations and 2 items for expectancies, using a 5 point agree-disagree, important-not important Likert scale, for response. A lower score on this scale indicates a higher perception that genomic medicine can be incorporated on a trial basis.

A related diffusion construct, "*observability*," is a characteristic of the innovation that refers to adopters being able to observe how an innovation works by watching someone else use it and acknowledge that it is beneficial. The more obvious the evidence or the more visible the positive results, the more likely it will be adopted by new users. As more primary care physicians are observed ordering genetic tests and referring patients to genetic specialists with positive results, the sooner other primary care physicians will adopt this innovation. Respondents in this study were asked if they agreed or disagreed with statements such as: "Most of my colleagues are assisting patients to make decisions regarding genetic services." Two expectation items and two expectancy items were used to measure observability, using a 5 point agree-disagree, important-not important Likert scale for response. A lower score for this scale indicates

a higher perception that colleagues of the respondents are adopting genomic medicine into their practice.

Moderator Variables

Norms, roles, and social networks are very important to the diffusion of an innovation. Diffusion theory proposes that the social networks or *social systems* through which innovations spread help govern the pace and extent of diffusion. According to Cain and Mittmann,[14] studies in the 1960's of physicians prescribing tetracycline demonstrated that doctors with more extensive social networks—those on hospital staffs, those in group practices, and those who consulted with other physicians—adopted the drug faster than doctors who were more socially isolated. Whether the respondents are involved in private or group practice was examined in this study as having a possible moderator effect upon the dependent and predictor variables.

Demographic Variables

Age, gender, ethnicity, year of graduation, medical school, specialty, and practice characteristics will be used as control variables in multivariate analysis and examined for possible differences or relationships between and among groups.

Definitions

Primary care physician (PCP) will be defined in this study as a medical practitioner who serves as a patient's first point of contact into the health care system and takes continuing responsibility for providing the patient's care.[15] Primary care encompasses internal medicine, pediatrics, obstetrics, gynecology, family practice, and general practice. Primary care includes health promotion, disease prevention, patient counseling and education, diagnosis and treatment of acute and chronic illnesses in a variety of health care settings.[15]

Private practice will be defined as medicine practiced independently by a physician. Group practice will be defined in this study as medicine practiced by a group of associated physicians[16]

Genomic medicine is defined as the use of DNA testing to enhance the quality of medical care including pre-symptomatic identification of predisposition to a disease, preventive intervention, selection of pharmacotherapy, and individual design of medical care based on genetic makeup.[17] Genomics has also been defined as the study of "all the functions and interactions of all the genes in the genome" including their interactions with environmental factors.[18(p1513)] Guttmacher & Collins [18,] distinguish between "genetics" and "genomics." "Genetics is the study of single genes and their effects. Genomics, a term coined only 15 years ago, is the study not just of single genes, but of the functions and interactions of all the genes in the genome."[18(1512)] The human genome is the full collection of genetic material in a human cell .[19] A glossary of genetic terms used in this manuscript is included in Appendix A.

CHAPTER II

BACKGROUND/LITERATURE REVIEW

This chapter is divided into three sections. The first section presents the history and background of the Human Genome Project. Finished ahead of schedule and under budget, this project is responsible for the evolution of genetics into genomic medicine. As providing genomic information and advice will become the domain of primary health care providers,[20] the second section of this literature review examines the role of primary care physicians and the barriers they face in the new genomic revolution. The third section illustrates the Diffusion of Innovations theory and describes some of its uses in the field of health care.

Genetics to Genomics

In the early 1950s, evidence from two experiments provided clues to the structure of deoxyribonucleic acid (DNA). Chemical analysis by Austrian-American biochemist Erwin Chargaff revealed the base components and proportions of DNA.[21] Next, English physicist Maurice Wilkins and English chemist Rosalind Franklin observed the regularly repeating structure of building blocks of DNA with the help of a newly developed imaging technique called x-ray crystallography. The images showed DNA was helical or shaped like a spiral (Figure 2). [21,22]

In 1953, American biochemists James Watson and Francis Crick worked together in England to build a three-dimensional replica of the DNA molecule using

ball-and-stick models (Figure 3).[21] Watson and Crick's proposed model showed how the DNA's two chains wound around each other, with paired bases inside. The two scientists suggested that DNA copied itself and enabled genetic information to flow from one generation to the next. [22] On April 25, 1953 in a one page scientific publication in the journal "Nature", Watson and Crick stated with simplicity, "It has not escaped our notice that the specific pairing we have postulated immediately suggests a possible copying mechanism for the genetic material."[23, (p737)]

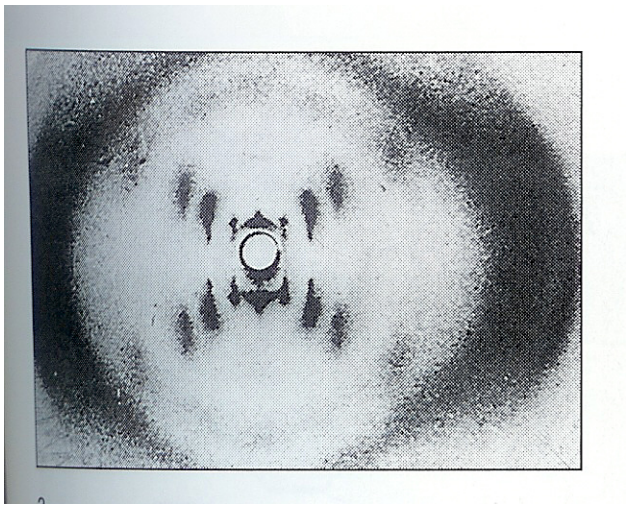


Figure 2 The X-ray Diffraction Pattern of DNA, Taken by Rosalind Franklin*

Although Watson and Crick's contributions still capture much of the public awareness, their breakthrough was one of many in an ongoing series of insights into the

* Courtesy of the Cold Spring Harbor Laboratory Archives. 2003.

fundamental design of life.[24] Gregor Mendel, a German botanist and Augustinian monk, formulated the basic laws that govern inherited characteristics less than a century earlier, and the 1940s saw the discovery of chromosomes as the sites that contain the genes.[21] Rosalind Franklin was a young woman when she contributed the x-ray diffraction data that would prove so pivotal to the elucidation of the structure of DNA. However, she did not share the Nobel Prize with Watson, Crick, and Wilkins because she died from ovarian cancer before the prize was awarded. It is speculated that she developed the cancer from her years of working with radioactive chemicals.[21]

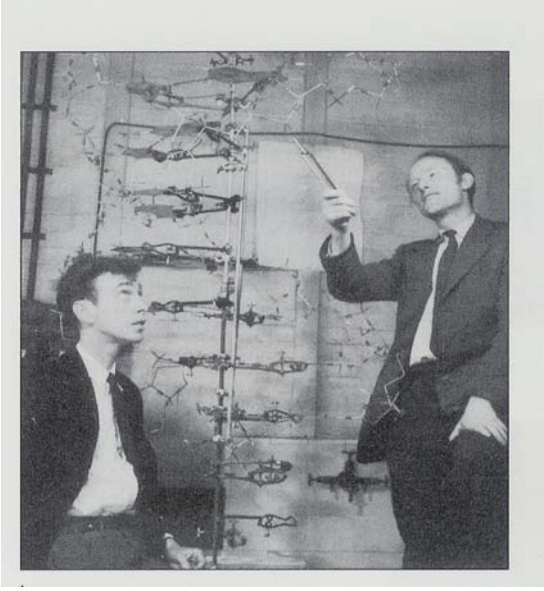


Figure 3 James Watson and Francis Crick with Their DNA Model[†]

The double helix discovered by Franklin and confirmed by Watson and Crick set the stage for sequencing the entire human genome "and turned genetics, the study of

[†] Image reprinted with permission. Copyright © 2002. Photo Researchers, Inc.

single genes, into genomics, the study of the interaction of all genes with one another and with their environments." [25 (p949)]

Increasing evidence of the importance of genetics in almost all diseases resulted in the development of the Human Genome Project in the mid-1980s.[26] The first serious discussion of sequencing the human genome was lead by Robert Sinsheimer, then chancellor of the University of California at Santa Cruz.[27] In 1986, Charles DeLisi of the U.S. Department of Energy (DOE) decided to begin funding research into genome mapping and sequencing.[27] In 1988, a special committee of the U.S. National Research Council of the U.S. National Academy of Sciences recommended the initiation of the Human Genome Project, calling for a 15-year project with funding of about $200 million a year.[27] The same year the National Institutes of Health (NIH), led by James Watson, joined the effort with the Department of Energy. When Watson departed in 1992, Francis Collins assumed the lead role at the NIH in 1993, and Michael Morgan at The Wellcome Trust (Great Britain) in 1992. Aristides Patrinos took the lead of the sequencing effort at the DOE in 1995. Recalling those early years, Collins, Morgan, and Patrinos[27] stated,

> The next several years were turbulent, as we learned 'on the job', made lots of mistakes, and experienced more than a few moments of great anxiety that the whole enterprise might fail; but ultimately, we watched the creativity, talent, and dedication of those involved in the public genome project surmount every obstacle and beat every deadline. We also realized that what we were learning had implications that extended beyond the human sequencing effort itself to the management of large-scale biology in general. [(p286)]

February 28, 2003 was the 50th anniversary of the discovery of the three-dimensional structure of DNA by James Watson and Francis Crick. In April 2003, a

"high-quality comprehensive sequence of the human genome" was completed 2 years ahead of time.[28 (p835)] More than 20 different major sequencing centers in six countries - China, France, Germany, Great Britain, Japan, and the United States - and hundreds of scientists participated in the project.[28]

The field of medical genetics traditionally covered a relatively limited range of single-gene disorders such as the ones listed in Table 1. Now it is known that of the 30,000 estimated chromosomal genes, about 1,000 carry disease-causing mutations. Moreover, Mendelian diseases that took large research teams years to identify in the past can now be identified in just a few weeks.[28] The Human Genome Project has identified hundreds of genes for an equal number of diseases such as cystic fibrosis, Huntington disease, breast and colon cancer, Retinoblastoma, Alzheimer's disease, and hemochromatosis. Specific chromosomal regions have even been identified for traits such as obesity, depression, violence, and homosexuality.[29]

The sequencing of the human genome and other recent achievements in genomics have provided an opportunity to advance our understanding of the role of

Table 1
Some of the Most Common Single-Gene Disorders[30,31,‡,32]

Condition	
Hypercholesterolemia	A dominantly inherited genetic condition that results in elevated LDL (low-density lipoprotein) cholesterol levels beginning at birth and resulting in myocardial infarctions (heart attacks) at an early age.
Diabetes, type I	Several different genes may act together along with other environmental and lifestyle factors to trigger diabetes. In type I diabetes, the beta cells of the pancreas produce little or no insulin, the hormone that allows glucose to enter body cells. Once glucose enters a cell, it is used as fuel. Without adequate insulin, glucose builds up in the bloodstream instead of going into the cells. The body is unable to use the high levels of glucose for energy.
Breast and ovarian cancer	Mutations in the BRCA1 and BRCA2 genes are associated with an increased risk of breast and ovarian cancer.
Down Syndrome	A chromosomal condition causing mental retardation and characteristic facial features. Other complications may include congenital heart disease and childhood leukemia.
Fragile X Mental Retardation	An X-linked syndrome that is the most common genetically-inherited form of mental retardation. Some children appear normal in infancy but develop intellectual disability and typical physical characteristics such as prominent ears during their lifetime. Mental impairment may range from mild to severe.
Sickle Cell Anemia	An autosomal recessive condition causing vasoocclusive events, resulting in pain, cerebrovascular complication, and renal dysfunction.
Cystic Fibrosis	An autosomal recessive condition causing progressive lung disease. Most affected patients also have pancreatic insufficiency.
Duchenne Muscular Dystrophy	An X-linked recessive condition causing progressive skeletal-muscle weakness and cardiomyopathy. Affected children are typically wheelchair-bound by the age of 12 years. Death is usually due to cardiomyopathy or respiratory failure.
Hemophilia A	A hereditary blood coagulation disorder caused by a deficient activity of plasma protein factor VIII, which affects the clotting property of blood.
Marfan Syndrome	An inherited collagen-based disorder characterized by long, thin arms, legs, and digits of the hands and feet. Ocular problems occur, and aortic heart disease most frequently is the cause of premature death.

‡ Adapted with permission from: Burke W. Genomic medicine: Genetic testing. *N Engl J Med*. 2002;347(21):1867-1875.

genetic factors in human health and disease, to allow more accurate explanation of non-genetic factors involved, and to apply this insight to the prevention, diagnosis and treatment of disease.[28]

Collins and colleagues[28] identified 3 large-scale strategies that will utilize the information from the Human Genome project to advance improvements in human health:

- Identification of genes and pathways with a role in health and disease, and determination of how they interact with environmental factors.
- Development, evaluation and application of genome-based diagnostic methods for the prediction of susceptibility to disease, the prediction of drug response, the early detection of illness and the accurate molecular classification of disease.
- Development and deployment of methods that catalyze the translation of genomic information into therapeutic advances.

Improvements in diagnostics and treatments resulting from these strategies are anticipated to change the future of medicine and will become the standard of care in primary practice.

Genomic Medicine in the Primary Care Practice

Primary care physicians (PCPs) are increasingly becoming the main point of entry for genetic services in many countries.[33] Many patients feel more comfortable revealing family history of unusual disease to their primary care practitioner rather than to an unfamiliar genetic specialist. This puts the PCP in a good position to offer prenatal counseling to his/her patients as well as advice about genetic procedures and available options.[33] However, most of the research examining PCPs' adoption of genetic medicine has uncovered four major barriers to the provision of genetic services: lack of knowledge, lack of referral skills, skepticism about the impact of genetics on primary care practice, and lack of information about available resources.

Lack of Knowledge

Kolb and colleagues[34] found the primary reason for underutilization of genetic services by primary care providers in a Texas community was lack of adequate genetics information. After administering a 16-hour basic genetic educational program supplemented by a 150-page course manual, several pamphlets on specific genetic conditions and a videotape that was developed by the Texas Department of Health, there was an increase in knowledge and change in attitudes of the primary care providers towards genetic services.

Focus groups with 26 general practitioners concluded that lack of genetic knowledge and referral skills were also a barrier to providing genetic services in Britain.[35] Although the physicians considered genetics as important and increasingly

relevant for primary care, they felt the added responsibilities of genetics will require new knowledge and skills beyond what they are currently practicing.

In 1933 M.T. Macklin[36] declared, "Medical genetics should be taught in medical school, in the final year of medicine…Such a course…would be of value from four distinct points of view: diagnosis, therapeutics, prevention, and public health." (p 16) Today the underemphasis on genetics in medical school is still being partially blamed for the knowledge gap. "As practicing clinicians, we are left to delve into the black box of genetics with our limited backgrounds and often with even less appreciation for the practical relevance of genetics to our daily practice." [37 (p6)] Dumont-Driscoll[37] feels the "scant 2 weeks" allocated in the medical curriculum has created an enormous need for physicians to seek additional education in a previously underemphasized area of medical education. (p6)

Williams[8] agrees that medical schools offering only an introductory genetics course during the first or second year compound PCPs' lack of genetic knowledge. This lack of training leaves PCPs undereducated in a field that is expanding so quickly that even genetic practitioners are hard-pressed to keep up. Medical schools have been slow to recognize the profound implications of genomics for clinical medicine.[25] "There is inadequate time in the crowded curriculum and insufficient faculty to teach the new science at the necessary cellular, clinical, epidemiological and ethical levels." [25 (p949)] Aggravating this knowledge gap will be those already in practice who will need continuing education in order to counsel patients regarding genetic testing and to help patients understand their specific health risks.[25]

In contrast, Korf [38] contends that an extensive knowledge of the science of genetics will not be necessary for PCPs to use genomic medicine in future practice. The author claims genetics can be used in day-to-day practice by employing only important concepts and skills of basic genetics and by using them wisely. As far as medical curriculum, education goals of students are not all the same. Some may be preparing for primary care while others for specialty practice, research or public health.[38] In agreement with Korf, Dumont-Driscoll[37] stresses, "The general concepts are critical, but the knowledge of their intricacies, although intellectually exciting, are no more necessary than our knowledge of computer hardware to successfully write a memo or significant expertise in technology to use our household electrical equipment."[(p7)] It will be a challenge to genetics educators to focus on material relevant to primary care practice as well as to persuade many physicians that they need only to focus on what is important.

Many PCPs will want to have a hand in determining the specific content of what they are taught about genetics.[20] However, with the rapid pace of new information, too narrow a focus will fall short of what PCPs are going to need to practice genomic medicine. A workforce in primary-level genetic services will require PCPs to "think genetically" with every patient.[39] While the PCP already evaluates each individual in the context of his or her family and community,[39] genomic medicine will move physicians away from viewing the human body as a machine towards realizing each individual's genome may respond differently to his or her environment.[37]

Skepticism about the Impact of Genomic Medicine

Skepticism about the impact of genetic discoveries on primary care practice could be a barrier to providing genetic services.[40] This doubt may cause genetic training to be a low priority for many primary care physicians.[41,42] Hayflick and colleagues[42] reported six percent of physicians in their study (examining primary care physicians' perceptions of genetic services) felt no need to include these additional services in their practice. Lack of effective screening technologies and therapies to reduce risk or prevent disease caused practitioners in Kumer and Gantley's[35] study to believe genetic advances had little relevance for their practice.

Rapidly evolving advances in genetic testing, pharmacogenetics, and gene based therapies have important implications for primary care physicians because they are mainly responsible for initial assessment of medical problems, disease prevention and long-term care of their patients.[43] However, the need for the development of additional genetic curriculum and continuing medical education might not seem urgent for the many PCPs who are skeptical about the relevance of genetics.[43]

A 1998 American Medical Association's survey found that 71% of patients with a possible family history of genetic disease would rely on their primary care physician for advice about genetic services.[44] Yet, thirty-nine percent of the PCPs responding to an American College of Obstetrics and Gynecology survey rated genetic issues as last among their priorities and two-thirds were not confident of their knowledge of genetics.[45]

Benítez-Bribiesca[29] complained that most of the success of genomic medicine has been with rare diseases that comprise only 5% of all our pathologies. Genes related to the most common diseases such as cancer, infections, arteriosclerosis, and mental disorders have yet to be conquered. Even though genetic discoveries have led to noteworthy advances in the ability to better understand and diagnose many rare genetic disorders, the lack of effective interventions suggests that genetic technologies will be slow to be adopted into primary care medicine.[46] Genetic screening measures historically have focused on reproductive issues, such as preconception screening for those at risk of being carriers of autosomal recessive diseases like Tay-Sachs disease and cystic fibrosis or prenatal diagnosis such as Down Syndrome. Newborn screening is generally mandated by state or federal government health policies and occurs outside the physician's responsibility.[47] Many PCPs do not yet feel much need to prepare for the kind of practice in which predispositional genetic testing for susceptibility to common adult disorders may become routine.

Pinksy and colleagues[47] complained,

> The impending 'genetic revolution' has been so over promoted by members of the scientific, corporate, medical, and political communities that it is tempting to dismiss the role of genetics in primary care entirely."(p47)

Chanock & Wacholder[48] confirmed two major gaps in the implementation of genomic medicine: "1) the chasm between existing genetic information and its clinical utility and 2) the translation of collected data into effective clinical practice." (p268) They questioned whether more data will mean more useful information for those health care practitioners who do not even know which genetic tests are available much less how to

interpret the results. Lack of a basic understanding of these expensive genetic tests will increase the chance that test results will be misinterpreted.[49] In addition to interpreting the tests, the patients will expect to know why a test is performed, what the consequences are, and its implication for clinical care.[48]

Lack of Referral Skills/ Information about Available Resources

There has been criticism that physicians are fed a rich daily diet of gene discovery and ethical dilemmas yet they lack basic information on the uses of genetic testing and the tenets of genetic counseling that will allow them to incorporate these concepts into their practice.[47] Primary care physicians (PCPs) may be more comfortable referring patients to a genetic clinic or specialist for testing and counseling than actually ordering the test themselves because they are unable to interpret the test results for their patients. The necessity to explain the medical and behavioral implications of a genetic test in the primary care setting will also require additional training.

Recognition by the primary care physician of the signs that indicate the need for a genetics referral is an essential part of providing the most accurate diagnosis, information about the risk of recurrence, and medical management for the patient and family.[50] Many PCPs are unfamiliar with the role of clinical geneticists (MDs primarily involved with diagnosis of genetic disorders and counseling of patients and their families).[36] The assumption is that a geneticist is a specialist working in research. Curtis[36] explains that misunderstandings of genetics and its applications are common and can differ depending on the background of the people involved. For example, molecular

geneticists that investigate cystic fibrosis may not be familiar with the role of bacterial genetics that investigates the resistance to antibiotics.

A genetics consultation is often much more time-consuming than most medical visits.[19] Obtaining a more extensive family history is an essential start in the assessment and management of genetic disorders. According to Elsas and Trepanier,[51] a complete family history includes collecting information on parents, siblings, daughters, sons, aunts, uncles, and grandparents, which is then formatted into a genetic pedigree. The history should also include information about all types of cancer in both the paternal and maternal lineages as well as ascertaining ancestry.

Another limitation to the typical "gatekeeper" system is not only that "primary care physicians must recognize the value of genetic intervention to write a referral, but the medical management department of the MCO [managed care organization] must agree and authorize the referral."[8 (p432)] For many PCPs and consumers, obtaining genetic subspecialty appointments is already difficult. Patients and consumers residing in smaller cities or rural areas have limited access to genetic specialists who are typically concentrated in academic medical centers in major cities.[52]

The increasing demand for genetic services will stretch the already limited number of specialists making it even more difficult for both urban and rural community physicians to consult with and refer patients to the appropriate specialist.[37] In an estimate of certified genetic specialists in the USA, Evans & Britt[44] found that there are approximately 3,000 physicians, nurses and counselors (not all of whom are engaged in clinical practice) serving a population of approximately 270 million people. Yet,

Pletcher and colleague's[53] study confirmed that reimbursement for geneticists and genetic counselors is just as poor now as it was 14 years ago despite the substantial professional time commitment involved providing genetic services.

Researchers question whether there is a need to train more clinical geneticists to provide for the increasing need for comprehensive genetic care or a need for geneticists to educate and train PCPs to incorporate genetics into their practice.[53] Despite reluctance by primary care practitioners to adopt these new responsibilities, "all branches of medicine, especially primary care, will be required to advise patients about genetic issues" [54(p1030)] As argued previously, clinical genetics departments will be unable to cope with the rise in demand of genetic referrals and consultations, given their existing workforce.[54]

Many primary care practitioners lack information about genetic services and options available to patients.[55] Not knowing which choices are available decreases their confidence to assess, counsel and refer patients to services offered by genetic clinics. Watson and colleagues [55] sought to determine if provision of printed materials alone was effective to disseminate new knowledge and implement guidelines successfully. One group of practitioners in this study was issued a tailored information pack while another group received an education session supplemented by the information pack. The second group fared no better in appropriate referral decisions than the group with the information pack alone. However, both groups fared better than the control group that did not receive either aid.

The trigger for a "need to know" more about clinical genetics has not really been elicited yet for most PCPs. An underlying cause for this possible denial is not knowing where or how to access the appropriate genetic services.[37] Lack of use of the services that are available may be tied to the PCP's skepticism of the value of genomic medicine. But Dumont-Driscoll[37] contends, "Believing that outcomes are not affected by the genetic information provided is inaccurate, inappropriately judgmental, and potentially a high risk for the patient and physician." (p9)

Ethical, Social, and Legal Issues

Barriers to implementation of genetic technology or skepticism about the clinical validity and utility of genomic medicine will no doubt hinder PCPs incorporating genomic medicine. Additionally, there will be many ethical, social, and legal issues that will need to be addressed. Apprehension about possible privacy breaches, stigmatization and discrimination may cause physicians and consumers alike to avoid genetic related services as well as participation in research.[56]

Discussions regarding the implications or applications of genomic medicine have not included the perspectives of the general population, especially the voices of racial or ethnic groups.[57] Historically in the United States, as well as in Europe, explanatory causes of particular diseases or social problems have been viewed as arising out of genetic material. For example, the eugenics movement in the United States in the early 1900s proposed solutions to social problems such as poverty by encouraging fertility among the upper and middle classes and discouraging childbearing among those from

lower socioeconomic groups.[57] Another example is the period during which Nazi war criminals of the 1930s and 1940s singled out Jewish, gypsies, and homosexuals on the grounds that they were genetically inferior.[57]

Pletcher and colleague's[53] survey data indicated that Hispanic and African American practitioners are extremely underrepresented as geneticists. The authors felt the ethnocultural changes in the U.S. suggest a need for a more racially and ethnically diverse genetic workforce as well as multicultural training in genetic residency programs.

Although it has been determined that there is no basis in the genetic code for race,[58] physicians need to be aware of the availability of population-based genetic screening such as for Tay-Sachs disease in Ashkenazi Jewish individuals or hemoglobin disorders in those of African, Mediterranean, or Asian descent.[38] As Lin-Fu and Lloyd-Puryear[59] stressed, this type of genetic screening and counseling is intended "to give the population at risk the information needed to make informed choices and prepare for the outcome of their decision." (p284)

Pre-conception screening or prenatal diagnosis is usually offered when there is a family history of genetic disorder or inherited chromosomal abnormality, when a couple already has an affected child, or if the parents are comparatively old.[10] Prenatal diagnostics are performed during pregnancy. The only options available following a prenatal diagnosis of an inherited disease are to give birth to an affected child, or to terminate the pregnancy. An alternative to prenatal diagnosis, pre-implantation genetic diagnosis, is a much less intrusive and invasive procedure involving the selection and

implantation of healthy embryos rather than abortion of an affected fetus.[45] Geller and colleagues[60] found that primary care providers are much more likely than genetic specialists (~44% of PCPs vs. ~16% of medical geneticists) to express an opinion to the patient whether or not she should opt for prenatal diagnosis. However, preimplantation diagnosis is rarely suggested to women as an alternative option to prenatal diagnosis by most physicians because it makes the decision-making process more complex when having "to differentiate the 'acceptable' from the 'non-acceptable' in terms of gender or other characteristics" which may devalue genetic diversity.[45(p754)]

Adams and Cain[45] point out that the intent of both prenatal testing and pre-implantation genetic diagnosis is eugenic in that it aims to reduce the number of people with genetic disorders through the rejection of characteristics that may not be in and of themselves fatal nor cause severe suffering in childhood. In defense, Kitcher[61] feels that it is with the best intentions that physicians or parents justify prenatal testing and termination of a pregnancy if it would prevent the birth of a child whose life would be brief and agonizing. However, many PCPs as well as other groups worry that it would be very easy to move away from trying to avoid human suffering to attempting to implement full-blown eugenics.

By the year 2010, it is expected that predictive genetic tests will be available for as many as a dozen common conditions, allowing individuals who wish to know this information to learn their individual susceptibilities and to take steps to reduce those risks for which interventions are or will be available.[5] But access to expensive new

genetic tests will be difficult for primary care patients, especially for Medicaid recipients and the uninsured.[45]

The discovery of genetic predisposition to chronic and inherited disease has set up the potential for discrimination in everything from who is marriageable to who is insurable. Stigmatizing groups that have been labeled high risk may not only result in their inability to obtain health insurance but also to obtain certain jobs.[44] Patients who are eligible for health care services under Medicaid may not experience barriers to genetic services for financial reasons to the degree encountered by those who are employed in low paying jobs with little or no health care benefits or those with private insurance. And if insurers are allowed to use genetic information in adjusting premiums, the burden of paying large if not impossible sums to obtain health care coverage will afflict numerous Americans who never thought of this as their problem.[62]

According to Cunningham,[62] when obtaining consent for any genetic tests, risks and benefits must be discussed, including the potential for false-positive and false-negative test results and the potential effect of these findings on self-image, family relationships, employment, and insurance coverage, as well as the physical and emotional burden of the disorder. As if this would not take up enough consultation time, the patient also needs to understand the difference between screening tests, which could place a person in a high-risk group, and diagnostic tests, which dictate treatments.[62]

One of the most complex issues accompanying the evolution of genetic sciences relates to how the results of genetic tests are to be used. Federal legislation has been proposed to define the rights of insurers and consumers; no significant efforts, however,

have been passed directly relating to this issue.[1] Consumers believe that allowing insurers access to genetic information would violate rights of privacy, prevent patients from getting needed help, and lead to widespread discrimination against applicants.[63] However, Pokorski and colleagues[64] make a good case that lack of data regarding the effectiveness of preventive treatment or interventions for patients with a genetic predisposition is consistent with other circumstances in which insurers feel compelled to deny coverage. Insurance companies have a natural interest in this data because assessment of risk is an essential factor in profitability.[64]

Another concern for PCPs is the psychological and emotional impact that positive test results can have on some patients. Especially patients with a family history of a disorder such as Huntington's Disease (also known as Huntington's Chorea).[10] Affected individuals with this disorder undergo gradual neural degeneration until they die. This lethal disorder is particularly tragic because it has such a late onset, typically at about age 40. By that time, the affected individual may have produced a family. Each child has a 50% probability of inheriting the disorder and transmitting it to his/her children.[32]

In families with Huntington's disease, individuals are often painfully aware of their risk, having lived for years with mixed hope and dread. Many people desire genetic information, even after they are made to understand its impact. In many cases, the family doctor is the most appropriate person to do the counseling because he/she knows the family and its attitudes and background better than an outside consultant. However, the PCP may have neither the genetic knowledge nor the time. Moreover,

some cases may be so complex or require such specialized tests that the services of a professional genetic counselor are needed. When the patient's decision to undergo testing reflects a desire to move forward in some way, and to end a long-standing struggle with anxiety and uncertainty, having to disclose a positive test can be very emotional.[10]

Evans and Britt[44] brought up a potential budgetary conflict between two federal health efforts, the Human Genome Project and the Healthy People 2010. The authors point out that the "massive wishlist" of the Healthy People 2010 revolves around low-cost solutions to public health issues such as increasing women's folic acid intake to prevent neural tube defects. But nowhere in this lengthy document does it push for access by the general population of women to costly genetic innovations such as genetic screening for breast or ovarian cancer.[44] Genetics plays a role in approximately half of the ten leading health indicators selected as public health issues in the Healthy People 2010 document.[65] Of the total 467 objectives, only one deals specifically with genetics (newborn screening).

According to Grosse and Teutsch,[66] advances in human genetics will require systematic assessments as to their rational translation into public health policy and practice. They further explain that the prevention effectiveness of a public health strategy is decided based on whether it is considered more effective, less costly, and poses no risks of harm. However, they found it to be more common that a public health strategy that is superior on one or more criteria usually ranks poorly on another.

Companies that hold patents for genetic tests in order to offer them for profit will no doubt keep the costs of these tests high. It was agreed that all the data produced from the Human Genome Project would be "freely available and in the public domain, in order to encourage research and development and to maximize its benefits to society."[67] (p29) Yet three million genome-related patent applications have already been filed. While it will take years to interpret much of the data detected by the Human Genome Project, especially with respect to common diseases, the physicians are concerned that commercial interests are already capitalizing on certain aspects of genomic medicine. To maximize revenues, many genetic centers and laboratories could deviate from nondirective counseling by prescribing unnecessary tests.[60]

Primary care providers get to know their patients well from both the medical and personal perspective. Because primary health care providers are often the first to be asked about hereditary disorders, he or she is in a unique position to direct the use of genetic technology for decision-making. But physicians are trained primarily to treat disease once it occurs.[68] Improved predictive capabilities provided by the Human Genome Project will benefit individualized risk assessment when relatively minor changes early in life may postpone or prevent the onset of disease.[69] However, complexities such as updating genetic knowledge, allotting more time for genetic services, and addressing the ethical, social, and legal issues of genomic medicine may impede the adoption of this innovation into primary care.

The Diffusion of Innovations in Health Care

Everett Rogers defines diffusion as the process by which an innovation is communicated through certain channels over time among the members of a social system.[11] There are four elements that are present in the diffusion of innovation process: the innovation, the communication channels, time, and the characteristics of the social system.

The *innovation* is an idea, practice, or object that is perceived as new by an individual or other unit of adoption. The characteristics of an innovation are strong indicators of the rate of diffusion and adoption.[70] The characteristics are divided into five categories that affect the rate at which an innovation gets adopted.[71]

Relative Advantage – Is the innovation perceived to be better than the status quo?

Compatibility – How does the innovation fit with people's past experiences and present needs? Does it require a change in existing values?

Complexity – How difficult is the innovation to understand and apply?

Trialability – Can people "try out" the innovation first? Or must they commit to it all at once?

Observability – How visible are the results of using it? If people adopt it, can the difference be recognized by others?

The *communication channel* is the path of information flow between and among individuals – the means and medium of communication. Mass media and interpersonal channels are two types of channels typically identified.[70] New information related to health is diffused through professional peer-reviewed journals and face-to-face at

meetings. On the surface, the limited channels of communication – journals, professional meetings – make it appear that the communication process would be easy. But the multiple layers of professional specialty groups within the health field, each with their own journals can actually slow down the diffusion process.[72]

The concept of *time* in the diffusion process is noticeable in three distinct areas: the innovation-decision process or the course taken from initial awareness of the innovation to its full adoption, the innovativeness of the individual or other unit of adoption, and the rate of an innovation's adoption within a system.

A *social system* is defined as a set of interrelated units that are engaged in joint problem solving to accomplish a common goal.[11] The social structure of a system is the hierarchical arrangement of its members. The social systems in health and medicine are hierarchical.[73] For example, medicine tends to have more political clout than does public health. Also, more attention is drawn to research findings in medicine than in other health fields such as nursing, which is considered a lower-status field.[72] The social system directs the path of diffusion and the behaviors of the system's individual members dictate the rate and volume of its flow.[70]

Brief History of the Diffusion of Innovations Theory

The original diffusion research was done in 1903 by the French sociologist Gabriel Tarde. Tarde plotted the original S-shaped diffusion curve where the rate of adoption increases slowly at first, then rises rapidly, and finally slows down and levels off (Figure 4). In his *Laws of Imitation,* Gabriel Tarde argued that proximity led to imitation.[73] In the 1920's a group of British and German-Austrian anthropologists used

"diffusionism" to explain how social change in a given society was a result of the introduction of innovations from another society.[11 (p41)]

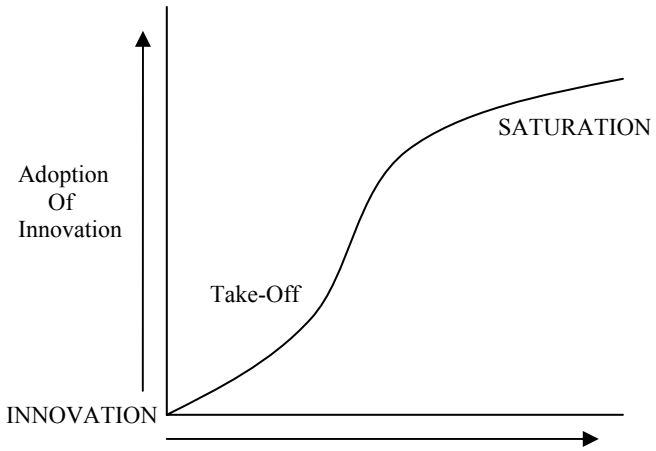


Figure 4 The Innovation Adoption Curve[71]

Early applications of the Diffusion of Innovations theory involved research to understand how agricultural techniques were spread among farmers.[73,74] In the 1940's, two sociologists, Bryce Ryan and Neal Gross published a study of the diffusion of hybrid corn seed among Iowa farmers, renewing interest in the diffusion of innovation S-curve. The rate of adoption curve was similar to the S-shaped diffusion curve graphed by Tarde.[75] Ryan and Gross classified the segments of farmers in relation to the amount of time it took them to adopt the innovation.[75] The five segments of farmers who adopted the hybrid corn seed, or adopter categories were: 1) innovators, 2) early adopters, 3) early majority, 4) late majority, and 5) laggards (Figure 5).

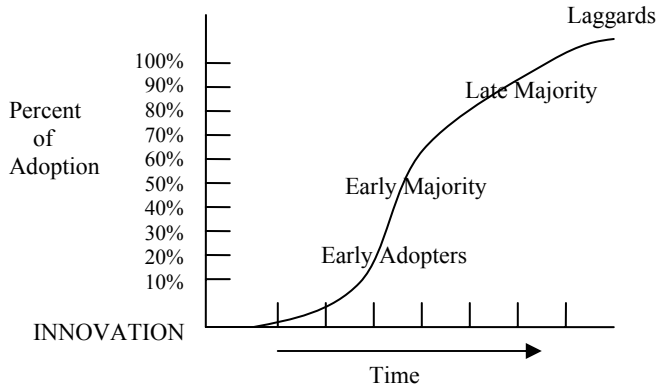


Figure 5 Adopter Categories of Innovation S-Curve[73]

In 1953, H.G. Barnett suggested that the likelihood of adoption of a new practice depends on the relative gain in satisfaction over current practices.[73] In 1963, Katz, Levin and Hamilton defined diffusion as "a sociological process characterized as the acceptance over time of some specific idea or practice by individuals, groups, or other adopting units, linked to specific channels of communication, to social structure, and to a given system of values or culture."[73 (p 166)] In 1983, Rogers[11] proposed that innovations could be characterized by their relative advantage over alternative products or behaviors, their compatibility with existing values, their complexity, trialability and their observability.

Diffusion of Innovations in Health Care

Studies using the Diffusion of Innovations theory to examine the perceptions of primary care physicians toward the innovation of genomic medicine have not been published. Therefore applications of Diffusion of Innovations theory to health care are

reviewed in this section. In the field of health care, the theory has been used to study family planning behaviors, the use of medical tests and technologies, and the use of new pharmaceutical agents.[73,74] Interventions for smoking cessation, safe sexual practices, dissemination of community-based cholesterol and diabetes screening are just a few ways earlier applications of the Diffusion of Innovations theory have given way to its extensive adoption by health education and health promotion.[73,74]

Svenkerud and Singhal[76] investigated the effectiveness of specific concepts from Diffusion of Innovations (DOI) in an effort to outreach culturally unique populations at high risk for HIV/AIDS in Bangkok, Thailand. They found that only 2 concepts of DOI were utilized consistly by the HIV/AIDS programs – communication channels and innovation attributes. Mass media strategies such as radio, television, and films were used as an initial information-spreading source. Given the sensitive nature of HIV/AIDS topics, interpersonal channels of communication were more effective in opening lines of communications and building trust between the outreach workers and the target audience. The programs were also sensitive to how the target audience perceived the attributes of the innovative HIV/AIDS programs. For example, the free services and colorful vests that were awarded to participants were viewed as a relative advantage. The programs also tried to be compatible with the day-to-day lifestyles and values of their clients.

Ash and colleagues[77] used the Diffusion of Innovations theory to study adoption and implementation of electronically entering physicians' orders for patient care (Physician Order Entry) in 4 hospital sites. This qualitative study collected data through

participant observation, focus groups, and formal face-to-face interviews. Patterns that fit the DOI Theory were then analyzed. The innovation attributes that were evaluated found that Physician Order Entry (POE) has some relative advantage over handwritten orders but may not be very compatible with the workflow or needs of users unless the hospital system values technology. Physician Order Entry was rated highly complex compared to handwritten orders but low on trialability since the hospitals tended to commit to POE without experimenting first. Since opinion leaders tended to sway the decision to implement POE, observability was rated high. Additional time required was resented by some users who perceived the chief benefits to be for someone other than themselves. According to the authors, benefits of POE depend on a critical mass of users sharing the order information. The larger the number of users, the faster POE diffused through the hospital social systems.

Pearcey and Draper[78] explored the factors associated with non-utilization of research-based preoperative information given to patients in a hospital ward. Roger's Diffusion of Innovations model of stages in the innovation-decision process supplied the framework for both data collection and analysis (Figure 6). In the *knowledge* stage, the study found that patients were not given adequate preoperative information. There was also a problem of not documenting important comments from patients or other details that might be important to recall later. Staff worked under the assumption that patients were already being given adequate preoperative information.

During the *persuasion* stage the researchers discussed the perceived characteristics of the innovation of a preoperative information protocol with the staff.

During the *decision* stage, staff agreed to write the protocol based on research provided by the researchers and use the innovation on a trial basis. This study did not make it to the *implementation* stage. It was postulated by the authors that the preoperative information protocol was not utilized because the more influential staff in the hospital did not feel the innovation was useful. However, it was felt that the Diffusion of Innovations framework could be useful in discovering why this type of protocol is rejected or adopted by other hospitals.

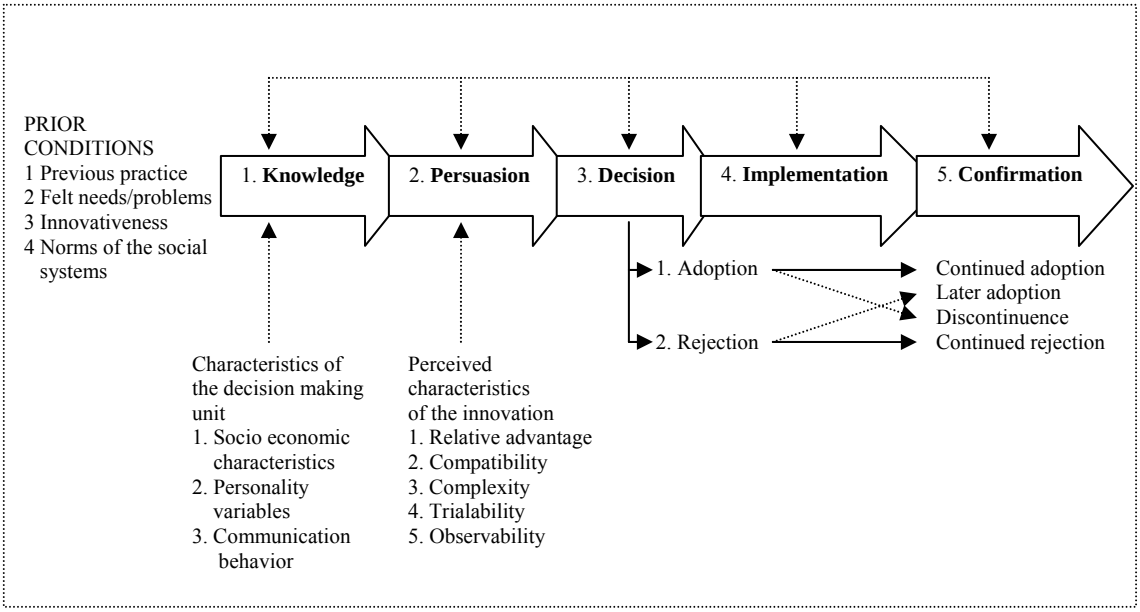


Figure 6 A Model of Stages in the Innovation-Decision Process[78]

Landrum[79] explored the theoretical perspectives of how people adopt innovations using the Standard Of Care: Skin Integrity protocol used in a medical intensive care unit at a large urban hospital. The information was used to plan campaigns to introduce innovations as a process rather than an outcome in medical settings. Landrum found the

rate at which nurses adopt and use research findings has a lag time of 8 to 30 years. The author states:

> In today's turbulent health care environment, the general public, administrators of health care agencies, and economics dictate that we decrease the lag time needed to adopt innovation into daily practice. Failing to do so can result in a loss of agency income and a loss of nursing jobs.[(p194)]

Research by Mary Moore, Dean of Library and Information Resources at the State University of Arkansas showed that the adoption of medical innovations such as telemedicine is influenced by factors such as socio-economics, demographic, psychological and communication related characteristics.[80] An interactive video (MEDNET) was implemented in 2 remote sites in west Texas where no large cities or medical centers were located. The purpose of the interactive video was to supply qualitative health care to underserved patients and to reduce the professional isolation of local physicians.[80]

The user characteristics of the sites inspired Dr. Moore to investigate physicians' adoption of medical innovations. While one remote site showed successful results, the other was an expensive failure. Users from the successful site turned out to be recent medical graduates, who were assertive and energetic, displaying a humble and altruistic attitude but self-confident at the same time. According to Moore,[80] these early adopters, acting as true opinion leaders, would be characterized by Roger's diffusion theory as having greater intelligence, abstraction and rationality.

According to Ference,[73] the early adopters of Coleman, Katz, and Menzell's 1966 study of the diffusion of a new antibiotic were also acting as opinion leaders:

> …they were younger (and perhaps more innovative), heavier prescribers of other drugs (perhaps from greater exposure to 'detailing'), more likely to be specialists (high status), and to have attended north-central medical schools, to read professional journals, attend conferences, and visit other institutions (better educated, greater exposure to external sources of communication.) [(p166)]

Identification of the two major breast cancer genes, BRCA1 and BRCA2 made testing for genetic susceptibility possible. Armstrong and colleagues[81] used components of the Diffusion of Innovations theory to describe the characteristics of women who were early adopters of BRCA1/2 testing. Variations in seeking BRCA1/2 testing after genetic counseling were also evaluated. The researchers concluded that the use of BRCA1/2 testing among women who were counseled is associated with innovative characteristics of the participant as well as the perceived compatibility of the test with personal values and needs. Attitudes about complexity and relative advantage of the tests were not associated with level of innovativeness.

Spellman[82] stated, "The revelations of genomics have fueled expectations that genetics will define human nature and that gene therapy will alter the natural history of disease."[(p665)] However, this author is cautious about these expectations when social determinants of health are still significant factors globally affecting medicine in the 21st century. In terms of who does or does not have access to genetic services, Evans and Britt[44] sense that advancement in genetic knowledge will bring a number of scenarios or patterns of diffusion, each with its own set of outcomes and implications. From past experience with diffusion of medical innovations such as prenatal diagnosis technology, the authors surmise that those who will benefit most from advances in genetic knowledge will probably be those who have the resources to do so.

Evans and Britt[44] observed that there has not been an emphasis on the diffusion of knowledge being generated by the Human Genome Project. According to Jones,[83] "new medical science knowledge usually kick-starts aggressive adoption by providers that perceive themselves as front-runners in their service areas. But this has not been the case with the new genetic technology."(p15) Emery and Hayflick[54] feel that specific elements of genetic medicine will require a gradual adoption and incorporation into primary care.

Given the complex nature of genetic knowledge and technologies, as well as the complexity involved in their adoption, understanding the diffusion characteristics of genomic medicine within a primary care environment will help address the potential needs of these physicians as genomic medicine is integrated into their practice. As most of the empirical research of PCPs' knowledge or attitudes about genomic medicine so far has been atheoretical, this study adds to the existing body of knowledge by examining the factors proposed by the Diffusion of Innovations theory that may influence PCPs' adoption of genomic medicine into their practice.

CHAPTER III

METHODS

The proposals for the pilot and final study were reviewed and approved by the Institutional Review Board of Texas A&M University (Appendix B). The purpose of the study is to assess whether and to what extent physicians' perceptions of genomic medicine as an innovation influence their likelihood of adopting this innovation into primary care.

Design

This study employed a survey design. An instrument to measure the perceptions of the characteristics of genomic medicine as an innovation and the likelihood of its adoption by primary care physicians was constructed and tested. Qualitative findings from a background study consisting of interviews with 6 primary care providers, 2 health educators, 1 geneticist, and 1 genetic counselor were used to inform the development of the instrument. A panel of experts reviewed the questions for content validity. A pilot study to test the questionnaire was conducted with a convenience sample of 50 primary care physicians in Texas. Internal consistency (Cronbach alpha reliability coefficient) and factorial structure of the instrument's scales were assessed during the pilot and final studies.

Qualitative Background Study

Nine interviewees from six sites in College Station, Texas were selected for the background study. The sites included a prenatal clinic, a family planning clinic, and a genetic counseling clinic, all located in the same community health center but without affiliation with each other. These three clinics serve all populations but most clients are funded primarily by Medicaid and federal Maternal and Child Health Bureau Title V funds. Interviews were also conducted at a fourth clinic site that provides service only to their health maintenance organization (HMO) clients. The fifth and sixth interview sites were Texas A&M University and the Texas A&M University Health Science Center.

Individuals from five professional groups participated in an interview process after informed consent was obtained. The interviewees included 2 family practice physicians, 2 nurse practitioners, 2 social workers (1 was also a genetic counselor), 2 health educators and 1 geneticist. The interviewees were chosen from referrals made by colleagues and other study participants. Interview sessions were typically from 30 minutes to 1 ½ hours in length. Three interview guides were developed; one for the family physicians, nurse practitioners, and social workers, one for geneticists, and another for the health educators (Appendix C). The interview guides were composed primarily of open-ended questions. Using a grounded theory approach elicited detailed information while allowing flexibility for probing questions and issues that study participants considered important. All interviews were tape recorded and transcribed verbatim. A content analysis of the transcripts informed the development of the instrument for this study.

Instrument

The principal investigator developed the pencil-and-paper, self-administered instrument used in this study. The questionnaire was brief (four-pages) in order to better fit a physician's busy time schedule and hopefully increase the historically low response rate for physician surveys. The questionnaire consisted of 35 questions to measure the perceived characteristics of the innovation of genomic medicine. There were also six demographic questions. Scales were designed to measure relative advantage, compatibility, complexity, trialability, and observability of genomic medicine. A scale to measure the likelihood of primary care physicians' adopting genomic medicine was also developed. A copy of the instrument is included in Appendix D.

Rates of diffusion and adoption depend to a large extent on how certain characteristics of the innovation interact with the targeted social system. The behaviors or characteristics of the system's individual members can also dictate the rate of adoption. Some individuals or "innovators" within the system are more influential than others in affecting the flow of diffusion and adoption.[70] While characteristics of adopters are crucial for understanding the adoption process, it was necessary to keep the questionnaire brief and only the characteristics of the innovation were measured in this study.

The construct "perceptions" is being understood in this study as "attitudes." Operationally, attitudes were measured through two dimensions: beliefs (outcome expectations) and values (outcome expectancies). Expectation and expectancy questions

were asked for each of the characteristics of the innovation of genomic medicine: relative advantage, compatibility, complexity, trialability, and observability.

Multiplying each expectation and corresponding expectancy scores and summing these products computed respondents' "perception" scores for each of the characteristics. For example, the score for the *relative advantage* scale is a result of the application of the formula below, where 1a through 1d are the items measuring the advantages of genomic medicine (beliefs) and 2a through 2d measure how important the perceived advantages of genomic medicine are to the respondent (values).

$$\Sigma\ (1_a * 2_a + 1_b * 2_b + 1_c * 2_c + 1_d * 2_d)$$

The four expectation items use a five point agree-disagree Likert scale and the four expectancy items (How important is it to you that…?), use a five point important-not important Likert scale, for response. A lower score for this scale indicates a stronger perception of the relative advantage of genomic medicine.

The eight items developed to measure *relative advantage* of genomic medicine are based on the "uses of clinically applicable gene tests" derived from GeneTests, a website funded by the National Institutes of Health, the Human Resource and Services Administration, and the Department Of Energy. The website is available at http://geneclinics.org. This scale achieved an internal consistency of .79 (n=21) in the pilot study and .73 (n=400) in the final study.

A factor analysis was conducted for the pilot and final studies to determine if perceived attribute items clustered as expected. For the analysis, the principle components method was used with a varimax rotation. The factor analysis for *relative advantage* revealed three major factors explaining 68.3% of the total variance (Table 2). All items that loaded on factor 1 assessed the expectations and expectancies of "predicting genetically inherited disorders." Factor two's items related to "pharmacogenomics" and factor 3 contained the expectation and expectancy of "preimplantation diagnosis."

Table 2

Final Study: Factor Loadings for Relative Advantage Items

N=400	Factor		
	1	2	3
Factor 1 – Predictive genomics			
I believe one of the advantages of genomic medicine is to…			
1b. Perform carrier testing for a possible autosomal recessive disorder before the onset of symptoms.	.766	--	--
1c. Supplement a family history in predicting the risk of a health individual developing a disease.	.775	--	--
How important is it for you to be able to…			
2a. Test whether an individual possesses a copy of a mutated gene for an autosomal recessive disorder before the onset of symptoms?	.649	--	--
2c. Supplement a family history in predicting the risk of a healthy individual developing a disease?	.750	--	--
Factor 2 – Pharmacogenomics			
I believe one of the advantages of genomic medicine is to…	--	.861	--
1d. Supplement knowledge of previous medical history in predicting which medications will be most effective for specific patients.			
How important is it for you to be able to…	--	.908	--
2d. Supplement previous knowledge of medical history in predicting which medications will be most effective for specific patients?			
Factor 3 – Preimplantation diagnosis			
I believe one of the advantages of genomic medicine is to…			
1a. Diagnose a genetic condition in an embryo before it is implanted instead of waiting and doing an ultrasound later in the pregnancy.	--	--	.835
How important is it for you to be able to…			
2a. Detect a genetic condition in an embryo before it is implanted?	--	--	.843
% of Variance	28.050	21.463	18.832
Cumulative %	28.050	49.513	68.346

Compatibility was conceptualized as a latent variable measured by 3 indicators, compatibility with *professional beliefs*, compatibility with *personal beliefs and values*, and compatibility with *current medical practice*. The outcome expectation questions for compatibility with *professional* and compatibility with *personal* beliefs and values were adapted from similar questions found in The University of Glasgow's e-mail questionnaire on ethical problems in medical genetics. This questionnaire is available at http://www.gla.ac.uk/departments/medical genetics/mbethicsq.htm. The outcome expectancy questions, developed by the principal investigator, were intended to measure how important it is for respondents that genomic medicine be compatible with their professional and personal beliefs and values, as well as with their current medical practice. The indicators "professional beliefs" and "personal values and beliefs" are each measured with two items for expectations, using a five point agree-disagree Likert scale and 1 item for expectancy, using a five point important-not important Likert scale. A lower score on these two scales indicate a stronger perception of the compatibility of genomic medicine with the professional beliefs and the personal beliefs and values of the primary care physicians.

The compatibility with *current medical practice* scale was informed by both the literature review and the qualitative background study which confirmed that many primary care providers lacked the time and skills to incorporate genetic counseling, more detailed family history, or genetic testing into their practice. The compatibility with "current medical practices" indicator is measured with three expectation items, using a five point agree-disagree Likert scale. The response "already incorporated" was added

to measure the number of respondents that have already incorporated certain tasks of genomic medicine into their practice. The one expectancy item uses a five point important-not important Likert scale, for response. A lower score for this scale indicates a stronger perception of the compatibility of genomic medicine with the primary care physicians' current medical practice.

The factor analysis showed that the compatibility indicators were measuring 4 components accounting for 74.1% of the variance (Table 3). All of the items for the compatibility with the *current medical practice* scale loaded on factor one. However, "How important is it to you for genomic medicine to be easily incorporated into your primary care practice?" had a small loading of .392. The items within factor 2 were measuring the professional and personal beliefs toward predictive testing. Factor 3 contained professional and personal beliefs toward terminating a pregnancy when the child would be severely affected. Items within factor 4 related to the importance of genomic medicine being consistent with professional and personal beliefs and values. Because the professional and personal belief items tended to cluster together in the factor analysis, these two indicators were combined to form one variable. Separately the internal consistency was low for "compatibility with professional beliefs" and "compatibility with personal beliefs" for both the pilot (.39, .57, n=21) and final studies (.35, .30, n=400). Combining the two indicators raised the internal consistency to .65 (n=400) for the new variable compatibility with *professional/personal beliefs and values*. The compatibility with *current medical practice* scale achieved an internal consistency of .80 (n=21) in the pilot study and .74 (n=400) in the final study.

Table 3

Final Study: Factor Loadings for Compatibility Items

N=400	Factor			
	1	2	3	4
Factor 1 – Ease of incorporating genomic medicine into current practice				
9. How important is it to you for genomic medicine to be easily incorporated into your primary care practice?	.392	--	--	--
10. Genetic counseling could easily be incorporated into my primary care practice.	.868	--	--	--
11. Taking a more detailed family history could easily be incorporated into my primary care practice.	.801	--	--	--
12. Genetic testing could easily be incorporated into my primary care practice.	.848	--	--	--
Factor 2 – Predictive testing beliefs and values				
4. Offering predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is consistent with my professional standards.	--	.916	--	--
6. Predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is compatible with my personal values.	--	.904	--	--
Factor 3 –Termination of pregnancy beliefs and values				
3. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my professional standards.	--	--	.893	--
5. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my personal values.	--	--	.927	--
Factor 4 – Consistency of genomic medicine with beliefs and values				
7. How important is it to you for genomic medicine to be consistent with your professional standards?	--	--	--	.853
8. How important is it to you for genomic medicine to be consistent with your personal values?	--	--	--	.868
% of Variance	23.053	17.782	17.746	15.543
Cumulative %	23.053	40.836	58.582	74.125

The *complexity* scale was also informed by the literature review and qualitative background study; both found that locating genetic services and staying updated on genomic medicine-related knowledge were barriers for primary care providers. This scale consists of two items for expectations and two items for expectancies, using a five point agree-disagree and easy-difficult Likert scale, for response. A higher score for this scale signifies a stronger perception of the complexity of genomic medicine. The Cronbach alpha for this scale was .62 (n=21) in the pilot study and .60 (n=400) in the final study. The factor analysis for the scale measuring complexity revealed two major factors accounting for 73.8% of the variance (Table 4). The two expectancy items for this scale fell within factor 1 and the 2 expectation items fell within factor two.

The *trialability* scale was designed to measure the extent to which genomic medicine can be experimented before a commitment to full incorporation is made. The items were informed by the literature review and qualitative background study. Item 16, "Genetic technologies, unlike other medical technologies, cannot be incorporated on a trial basis," was reverse-coded in both the pilot and final study. Trialability is measured with two items for expectations and two items for expectancies, using a five point agree-disagree, important-not important Likert scale, for response. A lower score on this scale indicates a stronger perception that genomic medicine can be incorporated on a trial basis.

Table 4

Final Study: Factor Loadings for Complexity Items

N=400	Factor	
	1	2
Factor 1 – Complexity of genomic medicine expectancies		
How important is it for you…		
14a. To be able to locate available genetic services without difficulty?	.891	--
14b. To easily stay updated on genomic medicine-related knowledge?	.882	--
Factor 2 – Complexity of genomic medicine expectations		
How easy or difficult is it for you to…		
13a. Locate available genetic services?	--	.739
13b. Stay updated on genomic medicine-related knowledge?	--	.861
% of Variance	41.134	41.134
Cumulative %	32.684	73.818

The four item scale achieved an internal consistency of .79 (n=21) in the pilot study. However, reverse-coded item 16 caused the reliability score to drop to .30 (n=400) in the final study. Item 16 was dropped from the final analysis, raising the internal consistency score to .65 (n=400). The factor analysis for the three item scale for trialability showed all items loading on the same component accounting for 47.2% of the variance (Table 5).

The *observability* scale, also informed by the literature review, was based on the degree to which the benefits of genomic medicine were being observed by the respondents. Two expectation items and two expectancy items were used to measure observability, using a five point agree-disagree, important-not important Likert scale for response. A lower score for this scale indicated a stronger perception that colleagues of the respondents are adopting genomic medicine into their practice. Reliability for this scale was .64 (n=21) for the pilot and .60 (n=400) for the final study. The factor analysis showed the observability scale loading on two components. Factor 1 contained the expectancy items and factor 2 contained the expectation items, accounting for 80% of the variance (Table 6).

The factor analysis for the scaled dependent variable "Likelihood of PCPs adopting genomic medicine" showed that all five items were measuring this component, accounting for 57% of the variance (Table 7). This scale, which achieved an internal consistency of .78 (n=21) in the pilot study and .81 (n=400) in the final study was based on the "uses of clinically applicable gene tests" derived from GeneTests (available at http://geneclinics.org). Adding the respondents' scores from the five items that question primary care physicians' likelihood of adopting genomic medicine computed the score for this scale. The scale used a five point likely-not likely Likert scale, for response. The response "I Already Am" was added to measure the number of respondents that are already using clinical applications of genomic medicine. A lower score indicated a stronger likelihood of adopting the innovation of genomic medicine.

Pilot Study

A pilot study was conducted to pretest the questionnaire with a convenience sample of 50 primary care physicians in Texas. The physicians were chosen among acquaintances and from the telephone directory for Bryan, College Station, San Antonio, and Austin, Texas. The survey was accompanied by a cover letter explaining the study

Table 5

Final Study: Factor Loadings for Trialability Items

N=400	Factor
	1
Factor 1 – Trialability of genomic medicine – expectations and expectancies	
15. Genetic services can gradually be incorporated into primary care practice.	.730
16. Genetic technologies, unlike other medical technologies, cannot be incorporated on a trial basis.	-.466
How important is it for you…	
17a. Be able to gradually incorporate genetic services into your practice	.795
17b. Incorporate technologies that you have tried first?	.711
% of Variance	47.202
Cumulative %	47.202

Table 6

Final Study: Factor Loadings for Observability Items

N=400	Factor	
	1	2
Factor 1 – Observability of genomic medicine expectancies		
Before you consider adopting genomic medicine into your practice, how important is it for you that your colleagues…		
19a. Adopt genetic testing into their practice?	.868	--
19b. Assist patients in making decisions regarding genetic services?	.852	--
Factor 2 – Observability of genomic medicine expectations		
Most of my colleagues are…		
18a. Adopting genetic testing into their practice.	--	.920
18b. Assisting patients to make decisions regarding genetic services.	--	.923
% of Variance	42.803	42.803
Cumulative %	37.227	80.030

Table 7

Final Study: Factor Loadings for PCPs Likelihood to Adopt Items

N=400	Factor
	1
Factor 1 – Likelihood to adopt genomic medicine	
For your patients, how likely are you to…	
20a. Order carrier testing for a possible autosomal recessive disorder?	.835
20b. Order a preimplantation diagnosis to check for genetic disease in an embryo?	.627
20c. Order a predictive test for risk of disease?	.797
20d. Provide pre-conception counseling?	.750
20e. Refer them for a genetic consultation?	.772
% of Variance	57.695
Cumulative %	57.695

and an informed consent form; both were previously approved by the Institutional Review Board of Texas A&M University (Appendix E). A self-addressed stamped envelope was included to facilitate return of the survey. A return fax number was given as an alternative to returning the completed survey by mail.

Twenty-one physicians, 18 male and 3 female, returned the questionnaire for a response rate of 42%. Their ages ranged from 32 to 70 years (Table 8). Sixteen of the respondents practice in a private setting and 5 in a group setting. The practices were located in Bexar, Brazos, Dewitt, Harris, and Travis counties (Table 9).

Table 10 shows the majority of respondents agreed that preimplantation diagnosis, carrier testing, using genomic medicine to supplement a family history in predicting disease risk, and supplementing knowledge of previous medical history to predict effective medication were all relative advantages of genomic medicine. Except for preimplantation diagnosis where responses were divided, the selected applications for genomic medicine were considered “extremely important” or “somewhat important” by more than half of the respondents.

A majority of the responding physicians feel that predictive testing for diseases for which there is no cure is consistent with both their professional and personal beliefs.

Table 8
Pilot Study: Selected Demographic Characteristics

Variable	No. (n=21)	%
Current age		
Under 40	4	19.0
40-54	11	52.4
55 and Over	6	28.6
Gender		
Male	18	85.7
Female	3	14.3
Ethnicity		
White	17	81.0
Black	0	0
Hispanic	0	0
Asian/Pacific Islander	4	19.0
Medical School		
In-state	14	66.7
Out-of-state	6	28.6
Out-of-country	1	4.7
Year of Graduation		
1975 and later	7	33.3
1976-1989	8	38.1
1990 and earlier	6	28.6

Table 9

Pilot Study: Professional Specialty of Physicians

Specialty	No. (n=21)	%
Internal Medicine	5	23.8
Pediatrics	3	14.3
Obstetrics and/or Gynecology	2	9.5
Family Medicine	9	42.9
General Practice	2	9.5

Pilot Study: Practice Setting of Physicians

Professional Practice	No. (n=21)	%
Private	16	76.2
Group	5	23.8

Pilot Study: County of Physicians' Practice

County	No. (n=21)	%
Bexar	9	42.9
Brazos	5	23.8
Dewitt	3	14.3
Harris	2	9.5
Travis	2	9.5

Table 10

Pilot Study: Percentage Distribution of Responses Regarding the Perceptions of the Relative Advantage of Genomic Medicine

1. I believe one of the advantages of genomic medicine is to… n=21	Strongly Agree	Agree	I'm not Sure	Disagree	Strongly Disagree
	%	%	%	%	%
a. Diagnose a genetic condition in an embryo before it is implanted instead of waiting and doing an ultrasound later in the pregnancy.	14.3	42.9	19.0	14.3	9.5
b. Perform carrier testing for a possible autosomal recessive disorder before the onset of symptoms.	38.1	52.4	9.5	--	--
c. Supplement a family history in predicting the risk of a healthy individual developing a disease.	28.6	61.9	9.5	--	--
d. Supplement knowledge of previous medical history in predicting which medications will be most effective for specific patients.	23.8	52.4	9.5	14.3	--

2. How important is it for you to be able to… n=21	Extremely Important	Somewhat Important	I'm not Sure	Not Very Important	Not Important At All
	%	%	%	%	%
a. Detect a genetic condition in an embryo before it is implanted?	19.0	23.8	9.5	28.6	19.0
b. Test whether an individual possesses a copy of a mutated gene for an autosomal recessive disorder before the onset of symptoms?	38.1	33.3	14.3	4.8	9.5
c. Supplement a family history in predicting the risk of a healthy individual developing a disease?	38.1	52.4	--	--	9.5
d. Supplement previous knowledge of medical history in predicting which medications will be most effective for specific patients?	38.1	52.4	4.8	--	4.8

Cronbach α .79 (8 items)

Less than half agreed that termination of pregnancy when there is a substantial risk the child would suffer from serious abnormalities is consistent with their professional or with their personal beliefs (Table 11).

Most of the physicians responded that it was important to be able to locate genetic services as well as stay updated on genomic medicine-related knowledge, but 33.4% and 52.4%, respectively, acknowledged that such tasks were either "somewhat" or "extremely" difficult for them (Table 12). The majority of respondents felt it was important to be able to gradually incorporate genetic services into their practice (Table 13). A majority of the physicians disagreed that their colleagues were adopting genetic testing into their practice. However, it was an important consideration before most respondents adopt genetic testing into their practice (Table 14).

Table 15 shows that none of the primary care physicians in the pilot study have already adopted any of the selected applications of genomic medicine into their practice. Most of them responded that they were likely to adopt all of the applications of genomic medicine except preimplantation diagnosis to check for disease in an embryo.

Table 11

Pilot Study: Percentage Distribution of Responses Regarding the Perceptions of Compatibility of Genomic Medicine

n=21	Strongly Agree	Agree	I'm not Sure	Disagree	Strongly Disagree
	%	%	%	%	%
3. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my professional beliefs.	14.3	33.3	14.3	23.8	14.3
4. Offering predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is consistent with my professional beliefs.	23.8	61.9	4.8	9.5	--
5. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is compatible with my personal values.	14.3	28.6	14.3	14.3	28.6
6. Predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is compatible with my personal values.	28.6	52.4	4.8	9.5	4.8

n=21	Extremely Important	Somewhat Important	I'm not Sure	Not Very Important	Not Important At All
	%	%	%	%	%
7. How important is it to you for genomic medicine to be consistent with your professional standards?	61.9	33.3	--	--	4.8
8. How important is it to you for genomic medicine to be compatible with your personal values?	52.4	33.3	--	9.5	4.8
9. How important is it to you for genomic medicine to be easily incorporated into your primary care practice?	28.6	57.1	4.8	--	9.5

Table 11 continued

n=21	Already Incorporated	Strongly Agree	Agree	Disagree	Strongly Disagree
	%	%	%	%	%
10. Genetic counseling could easily be incorporated into my primary care practice.	4.8	4.8	52.4	28.6	9.5
11. Taking a more detailed family history could easily be incorporated into my primary care practice.	--	9.5	71.4	14.3	4.8
12. Genetic testing could easily be incorporated into my practice.	--	9.5	42.9	38.1	9.5

Cronbach α

Professional beliefs .39 (3 items)
Personal beliefs & values .57 (3 items)
Current medical practices .80 (4 items)

Table 12

Pilot Study: Percentage Distribution of Responses Regarding the Perceptions of the Complexity of Genomic Medicine

13. How easy or difficult is it for you to…	n=21	Extremely Easy	Somewhat Easy	I'm not Sure	Somewhat Difficult	Extremely Difficult
		%	%	%	%	%
a. Locate available genetic services?		9.5	42.9	14.3	28.6	4.8
b. Stay updated on genomic-related knowledge?		--	33.3	14.3	42.9	9.5

14. How important is it for you…	n=21	Extremely Important	Somewhat Important	I'm not Sure	Not Very Important	Not Important At All
		%	%	%	%	%
a. To be able to locate available genetic services without difficulty?		38.1	57.1	--	--	4.8
b. To easily stay updated on genomic medicine-related knowledge?		28.6	61.9	--	4.8	4.8

Cronbach α .62 (4 items)

Table 13

Pilot Study: Percentage Distribution of Responses Regarding the Perceptions of the Trialability of Genomic Medicine

	n=21	Strongly Agree	Agree	I'm not Sure	Disagree	Strongly Disagree
		%	%	%	%	%
15. Genetic services can gradually be incorporated into primary care practice.		4.8	71.4	14.3	4.8	4.8
16. Genetic technologies, unlike other medical technologies, cannot be incorporated on a trial basis. (reverse-coded)		--	28.6	19.0	42.9	9.5
17. How important is it for you…	n=21	Extremely Important	Somewhat Important	I'm not Sure	Not Very Important	Not Important At All
		%	%	%	%	%
a. To be able to gradually incorporate genetic services into your practice?		14.3	57.1	14.3	4.8	9.5
b. Incorporate technologies that you have tried first?		9.5	57.1	23.8	--	9.5

Cronbach α .79 (4 items)

Table 14

Pilot Study: Percentage Distribution of Responses Regarding the Perceptions of the Observability of Genomic Medicine

18. Most of my colleagues are…	n=21	Strongly Agree	Agree	I'm not Sure	Disagree	Strongly Disagree
		%	%	%	%	%
a. Adopting genetic testing into their practice.		--	9.5	23.8	52.4	14.3
b. Assisting patients to make decisions regarding genetic services.		--	19.0	28.6	33.3	19.0

19. Before you consider adopting genomic medicine into your practice, how important is it for you that your colleagues…	n=21	Extremely Important	Somewhat Important	I'm not Sure	Not Very Important	Not Important At All
		%	%	%	%	%
a. Adopt genetic testing into their practice?		--	61.9	9.5	19.0	9.5
b. Assist patients in making decisions regarding genetic services?		14.3	57.1	9.5	19.0	--

Cronbach α .64 (4 items)

Table 15

Pilot Study: Percentage Distribution of Responses Regarding the Likelihood of PCPs Adopting Genomic Medicine

20. For your patients, how likely are you to… n=21	I Already Am	Extremely Likely	Somewhat Likely	Not Likely	Not Likely At All
	%	%	%	%	%
a. Order carrier testing for a possible autosomal recessive disorder?	--	14.3	66.7	4.8	14.3
b. Order a preimplantation diagnosis to check for genetic disease in an embryo?	--	9.5	23.8	28.6	38.1
c. Order a predictive test for risk of disease?	--	9.5	66.7	14.3	9.5
d. Provide pre-conception counseling?	--	23.8	33.3	19.0	23.8
e. Refer them for a genetic consultation?	--	52.4	38.1	4.8	4.8

Cronbach α .78 (5 items)

Final Study

The pilot study was conducted between March and April of 2003. After reviewing the internal consistency scores and factorial structure of the instrument's scales, the survey was not changed for the final study. The final study was conducted between April 15, 2003 and June 1, 2003. The 42% response rate for the pilot study lent support to the need to choose an adequate sample size for the final study that would still obtain a representative sample of the target population despite a low response rate.

Sample

The population of interest for this study is primary care physicians practicing in the state of Texas. A random stratified sample of primary care physicians (N=1350) was chosen from a sampling frame obtained from the Texas State Board of Medical Examiner's December 2002 directory of practitioners. The database was sorted into the target primary care specialties (internal medicine, obstetrics and gynecology, pediatrics, family practice, general practice). The obstetrics and gynecology group included physicians who specialized in obstetrics, gynecology, or both obstetrics and gynecology. The pediatric group included all subspecialties such as pediatric endrocrinology, pediatric hematology/oncology, pediatric radiology, pediatric allergy, and pediatric cardiology.

All records of physicians that were not active in practice or direct patient care, retired, or deceased were excluded from the sampling frame. Records that did not show Texas as the practice state also were eliminated. A proportional sample was randomly

selected from each stratum (internal medicine, obstetrics and gynecology, pediatrics, family practice, general practice) using a table of random numbers. The sampling fractions were combined to form an estimate for the population. For a population size of 21,306 licensed primary care physicians as reported by the Texas State Board of Medical Examiners (December 2002), a total sample size of 377 was needed to reach statistical representation.[84] The adjusted sampling frame of 17,445 active primary care physicians did not alter this figure. The random sample of N=1350 was based on a response rate of 30%, common for this population, which would obtain slightly more (405) than the required sample of 377, [1350(.30) = 405] (Table 16).

Table 16
Proportional Stratified Sample of Primary Care Physicians

PCPs	Population N %	To Be Mailed Out	Sample Size Needed
Family Practice	6,285 (30%)	405	121
General Practice	1,159 (5%)	67	20
*Obstetric/Gynecology	3,030 (14%)	189	57
Internal Medicine	6,827 (32%)	432	130
Pediatrics	4,005 (19%)	257	76
Total	21,306 (100%)	1,350	404 (~30%)

* Obstetricians and gynecologists were added to the Obstetric/Gynecology stratum due to low sample size needed. [Ob, 37 (<1%), Gyn, 201 (<1%]. Only a sample size of 1 and 4, respectively, was needed.

Design and Data Collection

This study consisted of a survey design. The sample was surveyed by mailed questionnaire in 3 waves. The first wave was a mail-out package containing a cover letter, questionnaire, informed consent form, and a stamped preaddressed envelope (Appendix F). All surveys were number-coded to allow for additional mail outs to non-responders. To ensure confidentiality, no names were used to identify respondents. Approximately 2 weeks after the first mail-out, reminder post cards were sent to those who had not responded. Due to the historically low response rate from physicians, a second mail-out package had been budgeted for the third wave to be sent out to the remaining non-responders approximately 5 weeks after the initial mail-out. In addition to the stamped preaddressed envelope, the respondents were given a return fax number in an effort to increase the response rate. The three mail-outs (first questionnaire packet, reminder postcards, second questionnaire packet) resulted in 379 completed surveys or a 28% response rate. Thirty-five surveys were returned because of unknown addresses or because the physician declined to participate. No differences were found between pilot study respondents and final study respondents. The pilot study data were merged with the final study's data increasing the sample size to 400 (379+21). Ten questionnaires arrived after the analyses were completed and will be included in future publications of this study.

Data Analysis

Data were analyzed with the assistance of the statistical software SPSS®, version 11.0. Descriptive statistics were utilized to assess distribution of responses and to assess patterns of skewness and kurtosis; correlational techniques were employed to study the zero-order relationships (their strength and direction) among variables. Multivariate techniques such as multiple regression and factor analysis were also used to examine interactions among variables, structure of the latent variable, and for comparisons of groups of adopters (those not adopting the innovation versus adopters), and/or demographic comparisons. Structural equation modeling techniques were also employed to test the proposed theoretical model and to assess patterns of prediction for different groups of respondents (private vs. group practice).

CHAPTER IV

RESULTS

Analyses

Relationships were examined among perceived relative advantage, perceived compatability, a latent variable measured with 2 indicators (compatibility with professional/personal beliefs and values, and compatibility with current medical practice), perceived complexity, perceived trialability, perceived observability, and likelihood of PCPs adopting genomic medicine. The social system (private or group professional practice) was tested for a possible moderating effect upon the dependent and predictor variables (Figure 7).

Descriptive statistics were utilized to assess distribution of responses and to assess patterns of normality (skewness: kurtosis); correlational techniques were employed to study the zero-order relationships (their strength and direction) among variables. Multiple regression and factor analysis were also used to examine interactions among variables, structure of the latent variable, and for comparisons of groups of adopters (those not adopting the innovation versus adopters), and/or demographic comparisons. Structural equation modeling techniques were also employed to test the conceptual model and to assess patterns of prediction for different groups of respondents (private vs. group practice).

To reduce moderate positive kurtosis (3.850), a square root transformation was applied to variable 1c – "I believe one of the advantages of genomic medicine is to

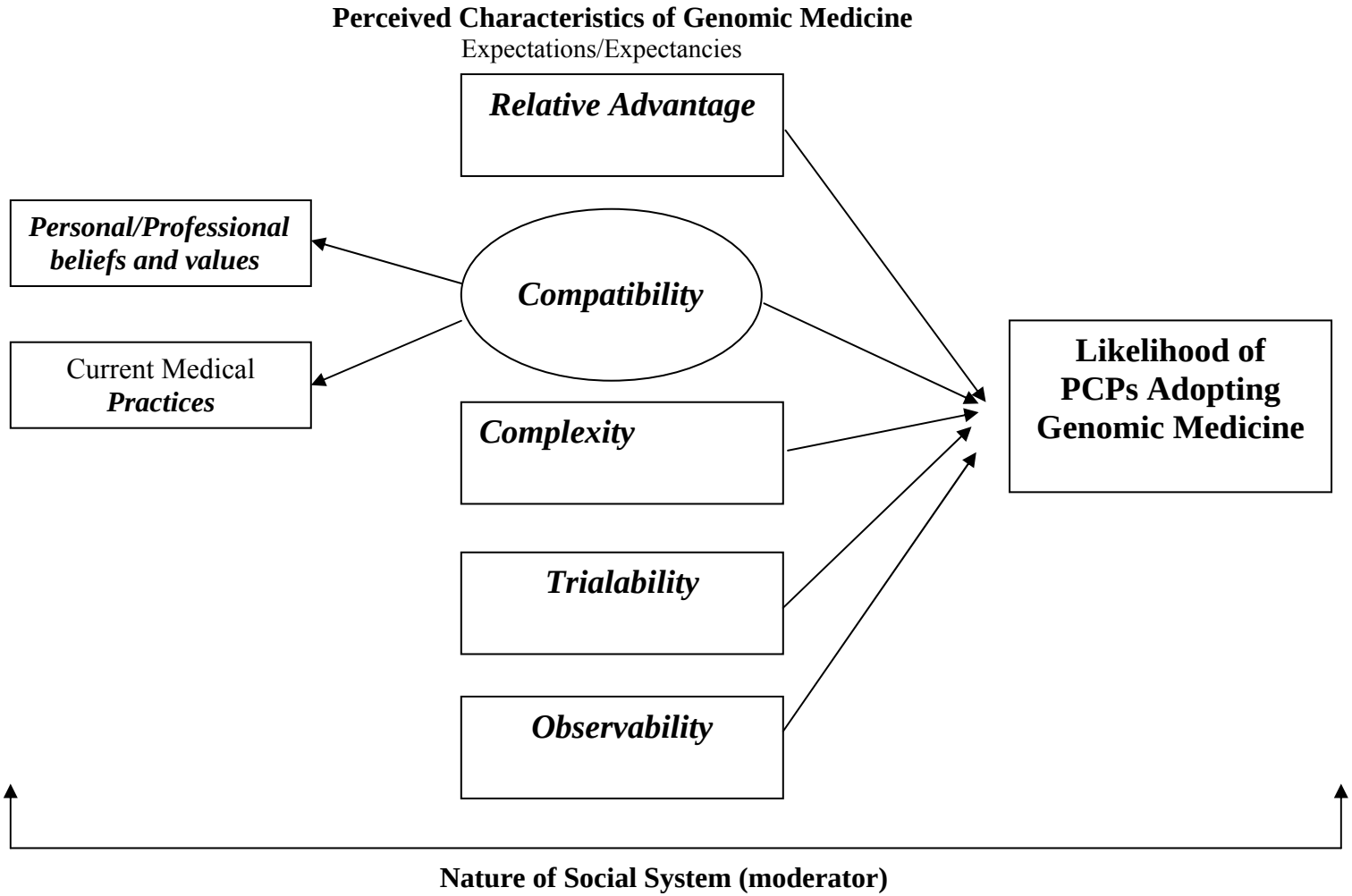


Figure 7 Model to Assess Likelihood of PCPs Adopting Genomic Medicine

supplement a family history in predicting the risk of a healthy individual developing a disease" – for all data analysis. A logarithmic transformation was used to reduce the substantial positive kurtosis of variable 7 (4.494) – "How important is it to you for genomic medicine to be consistent with your professional standards?"[85]

Missing Values

Missing data almost always generate analytical problems for which there is no ideal solutions.[85] Two questionnaires that were 25 to 50% incomplete were deleted from the study. Otherwise, responses to the questionnaires contained a small number of missing data points. Mean substitution was used on thirty-eight questionnaires that had 10% or less missing data points. The mean substitution procedure is considered conservative because "the mean for the distribution as a whole does not change and the researcher is not required to guess at missing values."[85 (p62)] Missing values were replaced with the means of the specific scale items prior to analysis. For example, cases with missing values in item 1a had the mean for 1a used for substitution.

Demographics

The final study's sample consisted of 400 primary care physicians whose ages ranged from 27 to 88. The mean age was 48.7 (SD = 11.76). The ages were collapsed into three groups (under 40, 40 to 50 years, over 50) for age comparisons. There were 286 (71.5%) males and 114 (28.5%) females that returned completed surveys (Table 17).

Table 17
Final Study: Selected Demographic Characteristics of Responding Physicians

Variable	No. (n=400)	%
Current Age		
Under 40	101	25.3
40-54	181	45.3
Over 55	118	29.5
Gender		
Male	286	71.5
Female	114	28.5
Ethnicity		
White	274	68.5
Black	11	2.8
Hispanic	42	10.5
Asian/Pacific Islander	67	16.8
Unknown	6	1.4
Medical School		
In-state	183	45.8
Out-of-state	99	24.7
Out-of-country	118	29.5
Year of Graduation		
1975 and later	129	32.3
1976-1989	158	39.5
1990 and earlier	113	28.3

The year of graduation from medical school ranged from 1940 to 2000. The median year was 1982. One hundred and eighty-three (45.8%) of the physicians attended a medical school in Texas, 99 (24.7%) attended an out-of-state medical school, and 118 (29.5%) graduated from a medical school in another country. One hundred and ninety-two (48.0%) of the physicians identified their medical practice as private and 151 (37.8%) as a group practice (Table 18). Fifty-seven (14.3%) of the responding physicians reported something other than private or group. In the final analysis, the physicians that reported they were affiliated with or worked in clinics, hospitals, or any other "group" atmosphere were aggregated with the group practice respondents.

The physicians were asked which professional specialty best described their practice. Ninety-five physicians (23.8%) reported their professional specialty as internal medicine, 96 (24%) pediatric, 57 (14.3%) obstetrics, gynecology, or obstetrics and gynecology, 95 (23.8%) family medicine, and 18 (4.5%) general practice. Thirty-nine (9.8%) reported their specialty as something other than what was listed (Table 19). The obstetrics, gynecology, and obstetrics and gynecology specialties were collapsed into one obstetrics/gynecology group for analysis.

A proportional sample was randomly selected from each stratum (internal medicine, obstetrics and gynecology, pediatrics, family practice, general practice) using a table of random numbers. The sampling fractions were combined to form an estimate for the population (see Table 16, page 74 in Methods). For a population size of 21,306 licensed primary care physicians as reported by the Texas State Board of Medical Examiners (December 2002), the study's sample size of 400 (379, final + 21, pilot) was

Table 18
Final Study: Practice Characteristics of Responding Physicians

Practice Setting	No. (n=400)	%
Private	192	48.0
Group	151	37.8
Other:	57	14.3

Table 19
Final Study: Professional Specialty of Responding Physicians

Specialty	No. (n=400)	%
Internal Medicine	95	23.8.
Pediatrics	96	24.0
Obstetrics and/or Gynecology	57	14.3
Family Medicine	95	23.8
General Practice	18	4.5
Other:	39	9.8

sufficient to reach statistical representation for the population of primary care physicians in Texas.[84] However, representation was not achieved for each individual specialty stratum, with the exception of the obstetrics/gynecology group (57 of 57 needed) and the pediatric group (96 of 76 needed) [Table 20].

Table 20

Sample Size Needed and Obtained to Reach Representation for Each Specialty Stratum

Specialty	Needed	Obtained
Internal Medicine	130	95
Pediatrics	76	96
Obstetrics and/or Gynecology	57	57
Family Medicine	121	95
General Practice	20	18
Other	--	39
Total	404	400

Of the 971 non-responders, 285 (29.4%) were female and 686 (70.6%) male. The ethnicities of the non-responders as reported by the Texas Board of Medical Practitioner's directory were 577 (59.0%) White, 59 (6.0%) Black, 137 (14.0%) Hispanic, 182 (19.0%) Asian, 1 (<1.0%) Indian, and 15 (2.0%) unknown. There was no significant difference between the gender of responders and nonresponders [$F(1,1369) = .000$, $p = .992$.] However, there was a difference between ethnicities of responders and nonresponders, [$F(1,1369) = 12.79$, $p < .001$.]

Descriptives for Criterion and Predictor Variables

The mean, standard deviation and possible range of scores for each of the study's predictor variables – Relative Advantage, Compatibility (as indicated by compatibility with professional/personal beliefs and compatibility with current medical practice), Complexity, Trialability, Observability – and for the criterion variable (Likelihood of PCPs Adopting Genomic Medicine) are shown in Table 21. A lower score for the predictor variable indicates a more positive perception of that characteristic of genomic medicine. A lower score for the criterion variable indicates a higher likelihood of adopting genomic medicine.

Table 21

Mean, Standard Deviation, and Possible Range for Predictor and Criterion Variables

Variables n=400	Mean	SD	Possible Range
Relative Advantage	21.24	13.06	(4-100)
Professional and Personal beliefs	18.13	13.13	(4-100)
Current Practices	23.43	16.64	(3-75)
Complexity	15.39	10.37	(2-50)
Trialability	10.55	7.31	(2-30)
Observability	23.79	12.82	(2-50)
Likelihood to Adopt	17.10	3.70	(5-25)

A series of one-way ANOVAs performed between each of the predictor variables and selected demographic variables showed no significant differences among age groups, gender, and year of graduation or practice setting.

Tables 22-24 provide descriptive statistics for each of the items in the scales. The mean and standard deviation for each item are for the sample as a whole. The scores were collapsed into three categories of responses for the percentages. The categories varied depending on the scale:

- Agree, not sure, disagree
- Important, not sure, not important
- Already incorporated, agree, disagree
- Easy, not sure, difficult
- Already incorporated, likely, not likely

Table 22 shows the majority of responders agreed that the selected clinical uses of genomic medicine were relative advantages. "Supplementing a family history in predicting the risk of a healthy individual developing a disease" had the highest percentage (91.0%). While the majority also agreed "diagnosing a genetic condition in an embryo before it is implanted" was an advantage of genomic medicine, this item had the lowest percentage of agreement (64.3%).

On the compatibility scale, 60.5% responded that it was consistent with their professional beliefs to terminate a pregnancy when there is a substantial risk of a serious mental or physical abnormality (Table 23). Only 56.5 % of the physicians, however, felt it was consistent with their personal beliefs. "I don't believe in abortions" was a written

Table 22

Final Study: Perceptions of the Relative Advantage of Genomic Medicine

Strongly Agree 1 / Agree 2 / I'm not Sure 3 / Disagree 4 / Strongly Disagree 5	Mean	St.Dev.	% who Agree	% Not Sure	% who Disagree
1. I believe one of the advantages of genomic genomic medicine is to…					
a. Diagnose a genetic condition in an embryo before it is implanted instead of waiting and doing an ultrasound later in the pregnancy.	2.47	1.28	64.3	13.3	22.5
b. Perform carrier testing for a possible autosomal recessive disorder before the onset of symptoms.	1.88	.95	88.8	3.0	8.3
c. Supplement a family history in predicting the risk of a healthy individual developing a disease.	1.86	.94	91.0	1.3	7.8
d. Supplement knowledge of previous medical history in predicting which medications will be most effective for specific patients.	2.11	1.18	81.0	2.0	17.0
Extremely Important 1 / Somewhat Important 2 / I'm not Sure 3 / Not Very Important 4 / Not Important At All 5	**Mean**	**St.Dev.**	**% Important**	**% Not Sure**	**% Not Important**
2. How important is it for you to be able to…					
a. Detect a genetic condition in an embryo before it is implanted?	2.62	1.38	55.3	18.8	26.0
b. Test whether an individual possesses a copy of a mutated gene for an autosomal recessive disorder before the onset of symptoms?	2.10	1.18	81.0	4.0	15.0
c. Supplement a family history in predicting the risk of a healthy individual developing a disease?	1.96	1.06	87.0	3.0	10.0
d. Supplement previous knowledge of medical history in predicting which medications will be most effective for specific patients?	2.06	1.20	82.3	3.0	14.8

Cronbach α .73 (8 items)

Table 23

Final Study: Perceptions of the Compatibility of Genomic Medicine

Strongly Agree 1 / Agree 2 / I'm not Sure 3 / Disagree 4 / Strongly Disagree 5	Mean	St.Dev.	% who Agree	% Not Sure	% who Disagree
3. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my professional beliefs.	2.49	1.34	60.5	16.0	23.5
4. Offering predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is consistent with my professional beliefs.	2.23	1.26	75.8	5.8	18.5
5. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is compatible with my personal values.	2.54	1.32	56.5	22.5	21.0
6. Predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is compatible with my personal values.	2.28	1.25	73.8	7.8	18.5

Extremely Important 1 / Somewhat Important 2 / I'm not Sure 3 / Not Very Important 4 / Not Important At All 5	Mean	St.Dev.	% Important	% Not Sure	% Not Important
7. How important is it to you for genomic medicine to be consistent with your professional standards?	1.69	.99	92.0	1.0	7.0
8. How important is it to you for genomic medicine to be compatible with your personal values?	2.07	1.31	82.5	1.5	16.0
9. How important is it to you for genomic medicine to be easily incorporated into your primary care practice?	2.29	1.31	75.8	3.8	20.5

Table 23 continued

Already Incorporated 1	Strongly Agree 2	Agree 3	Disagree 4	Strongly Disagree 5	Mean	St.Dev.	% Already Incorporated	% who Agree	% who Disagree
10. Genetic counseling could easily be incorporated into my primary care practice.					3.41	.93	3.3	48.3	48.5
11. Taking a more detailed family history could easily be incorporated into my primary care practice.					3.02	.88	5.8	70.3	24.0
12. Genetic testing could easily be incorporated into my practice.					3.41	.95	3.8	48.0	48.3

Cronbach α

Compatibility with professional/personal beliefs and values .65 (6 items)

Compatibiltiy with current medical practice .74 (4 items)

Table 24

Final Study: Perceptions of the Complexity of Genomic Medicine

Extremely Easy 1 / Somewhat Easy 2 / I'm not Sure 3 / Somewhat Difficult 4 / Extremely Difficult 5	Mean	St.Dev.	% Easy	% Not Sure	% Difficult
13. How easy or difficult is it for you to…					
a. Locate available genetic services?	3.00	1.50	50.0	11.8	38.3
b. Stay updated on genomic-related knowledge?	3.68	1.37	28.3	17.5	54.3
Extremely Important 1 / Somewhat Important 2 / I'm not Sure 3 / Not Very Important 4 / Not Important At All 5	**Mean**	**St.Dev.**	**% Important**	**% Not Sure**	**% Not Important**
14. How important is it for you…					
a. To be able to locate available genetic services without difficulty?	2.11	1.26	80.8	4.0	15.3
b. To easily stay updated on genomic medicine-related knowledge?	2.27	1.23	79.8	3.0	17.3

Cronbach α .60 (4 items)

comment on two of the surveys. Another respondent wrote, "I disagree with the ethics of physicians who perform abortions. However, I'd like to see genetics determined before sperm and egg unite as an embryo."

There was almost an even split between those who agreed that genetic counseling could easily be incorporated into primary care practice and those who disagreed (48.3% vs. 48.5%). Results for the item regarding genetic testing were similar (48% vs. 48.3%). However, most of the respondents felt they could incorporate a more detailed family history into their practice (70.3%). Over three percent are already doing some genetic counseling and 5.8% are already taking a more detailed family history. Almost four percent have already ordered some type of genetic testing for their patients. Half of the primary care physicians that responded to the survey found it easy to locate genetic services (Table 24). Not surprisingly, 54.3% found it difficult to stay updated on genomic-related knowledge, yet most (79.8%) felt it was important to do so.

The results of a one-way ANOVA revealed a significant difference between specialty groups and perceptions of the complexity of incorporating genomic medicine, [$F(5,394) = 16.481$, $p < .001$]. The obstetrics/gynecology specialty group obtained scores that were significantly lower than all of the other groups except pediatrics (Table 25). A lower score on this scale indicates that incorporating genomic medicine is perceived as less complex. This suggests that physicians from the obstetrics/gynecology and pediatric specialties compared to the family medicine, general practice, internal medicine, and "other" specialties do not perceive genomic medicine to be as complex.

Table 25

Group Mean and Standard Deviation for Complexity of Genomic Medicine Scale

Specialty	N	Mean	Std. Deviation
Internal Medicine	95	17.29	10.06
Pediatrics	96	10.75	7.77
Obstetrics/Gynecology	57	8.95	5.36
Family Medicine	95	19.71	10.89
General Practice	18	19.17	11.43
Other	39	19.43	12.37
Total	400	15.40	10.37

A Bonferroni post hoc analysis[85] confirmed that the obstetrics/gynecology and pediatric groups did not differ from each other but did significantly differ from the other four specialty groups. Cohen's d (the difference between the means, divided by the pooled standard deviation) was calculated to determine the size of this difference.[85] The magnitude in difference between the means of the obstetrics/gynecology group and the internal medicine, family medicine, general practice, and 'other' specialty group ranged from 0.49 to 1.27 standard deviations. The magnitude in difference between the means of the pediatric group and the internal medicine, family medicine, general practice, and 'other' specialty group ranged from 0.40 to 0.95 standard deviations.

Seventy four percent of the respondents agreed that genetic services could be gradually incorporated into primary care practice (Table 26). Over 60% felt it was important to gradually add genetic services and to try new technology before incorporating them into practice. Results shown in Table 27 indicate that a greater percentage of physicians disagree that their colleagues are adopting genetic testing or

Table 26

Final Study: Perceptions of the Trialability of Genomic Medicine

Strongly Agree 1 / Agree 2 / I'm not Sure 3 / Disagree 4 / Strongly Disagree 5	Mean	St.Dev.	% who Agree	% Not Sure	% who Disagree
15. Genetic services can gradually be incorporated into primary care practice.	2.50	1.19	74.3	2.8	23.0
Extremely Important 1 / Somewhat Important 2 / I'm not Sure 3 / Not Very Important 4 / Not Important At All 5	**Mean**	**St.Dev.**	**% Important**	**% Not Sure**	**% Not Important**
17. How important is it for you to…					
a. To be able to gradually incorporate genetic services into your practice?	2.85	1.42	61.3	6.3	32.5
b. Incorporate technologies that you have tried first?	2.74	1.32	64.8	5.0	30.3

Cronbach α .65 (3 items)

Table 27

Final Study: Perceptions of the Observability of Genomic Medicine

Strongly Agree 1	Agree 2	I'm not Sure 3	Disagree 4	Strongly Disagree 5	Mean	St.Dev.	% who Agree	% Not Sure	% who Disagree
18. Most of my colleagues are…									
a. Adopting genetic testing into their practice.					3.86	1.18	17.5	16.3	66.3
b. Assisting patients to make decisions regarding genetic services.					3.37	1.30	36.5	11.5	52.0
Extremely Important 1	Somewhat Important 2	I'm not Sure 3	Not Very Important 4	Not Important At All 5	Mean	St.Dev.	% Important	% Not Sure	% Not Important
19. Before you consider adopting genomic medicine into your practice, how important is it for you that your colleagues…									
a. Adopt genetic testing into their practice?					3.35	1.41	42.0	13.5	44.5
b. Assist patients in making decisions regarding genetic services?					3.08	1.45	51.5	10.3	38.3

Cronbach α .60 (4 items)

assisting patients to make decisions regarding genetic services (66.3% and 52%, respectively).

A between-subjects ANOVA was calculated with observability of genomic medicine as the dependent variable and specialty group (internal medicine, pediatrics, obstetrics/gynecology, family medicine, general practice, and other specialty) as the independent variables. There was a significant effect of specialty group, [F (5,394) = 4.39, p =. 001]. The Bonferroni post hoc analysis revealed that the internal medicine group (M = 28.0, SD = 13.18) and the obstetrics/gynecology group (M = 19.0, SD = 12.79) were significantly different from each other ($p<.001$, Cohen's d = 0.69) but not from the other groups. A lower score indicates a stronger perception of the observability of genomic medicine. In other words, the obstetrics/gynecology specialty group is observing their colleagues adopting genetic testing and assisting patients to make decisions regarding genetic services considerably more than the internal medicine specialty group. The pediatrics (M = 22.7, SD = 11.84), family medicine (M = 23.4, SD = 12.74), general practice (M = 20.6, SD = 13.46), and 'other' (M = 25.8, SD = 11.39) specialty groups did not differ from each other.

As shown in Table 28, the majority of the primary care physicians responded that they were likely to order carrier testing (55.5%), predictive testing (64.5%), and refer patients for a genetic consultation (80.5%). Three percent, 1.5%, and 2.5%, respectively, are already doing so. Almost 3% provide pre-conception counseling but the rest were split as to whether or not they are planning to incorporate this service (49% were likely to incorporate pre-conception counseling while 48.3% were not likely).

Table 28

Final Study: Likelihood of PCPs Adopting Genomic Medicine

I Already Am 1	Extremely Likely 2	Somewhat Likely 3	Not Likely 4	Not Likely At All 5	Mean	St.Dev.	% Already Incorporated	% Likely	% Not Likely
20. For your patients, how likely are you to…									
a. Order carrier testing for a possible autosomal recessive disorder?					3.35	.99	3.0	55.5	41.5
b. Order a preimplantation diagnosis to check for genetic disease in an embryo?					4.24	.90	--	20.5	79.5
c. Order a predictive test for risk of disease?					3.25	.94	1.5	64.5	34.0
d. Provide pre-conception counseling?					3.48	1.14	2.8	49.0	48.3
e. Refer them for a genetic consultation?					2.79	.89	2.5	80.5	17.0

Cronbach α .81 (5 items)

Eighty percent of the physicians responded that they were not likely to order preimplantation diagnosis and none were doing so at this time. Table 29 shows the number of physicians by specialty that have already incorporated some type of genetic service into their practice.

Table 29

Physicians by Specialty Group Already Incorporating a Genetic Service

Genetic Service n=400	Internal Medicine	Pediatrics	Obstetrics/ Gynecology	Family Medicine	General Practice	Other Specialty	Tot
Carrier testing	1	2	8	1	0	0	12
Preimplantation diagnosis	0	0	0	0	0	0	0
Predictive testing	1	1	3	1	0	0	6
Pre-conception counseling	1	3	6	1	0	0	11
Genetic referrals	1	2	7	0	0	0	10
Total	4	8	24	3	0	0	39

A between-subjects ANOVA showed a significant effect of specialty group on Likelihood of PCPs Adopting Genomic Medicine, [$F(5, 394) = 16.32$, $p<.001$]. The obstetric/gynecology specialty scored significantly lower on the "Likelihood of PCPs Adopting Genomic Medicine" scale than the other groups. A lower score on this scale indicates that this group is more likely to adopt genomic medicine than the other primary

care specialities (Table 30). The mean-level difference between the obstetric and gynecology group and the other groups ranged from 0.53 to 1.31 standard deviations.

Table 30

Mean and Standard Deviation for Likelihood of PCPs Adopting Genomic Medicine

Specialty	N	Mean	Std. Deviation
Internal Medicine	95	18.27	3.62
Pediatrics	96	16.56	2.71
OB/GYN	57	13.84	3.10
Family Medicine	95	17.53	3.70
General Practice	18	17.78	3.41
Other specialty	39	19.10	3.94
Total	400	17.11	3.70

A lower score indicates a stronger likelihood of adopting genomic medicine.
Possible range = 5-25

Test for Linearity and Multicollinearity

Pearson correlation measures the strength and direction of the linear relationship between the predictor and criterion variables.[85] Most of the relationships were significant at the .05 and .01 level. Table 31 shows that the criterion variable, Likelihood of PCPs Adopting Genomic Medicine, was linearly related to all of the variables in the model with the exception of the proposed moderator variable, Professional Practice (private or group practice) (r=.06). Its strongest association was with Complexity (r=.46, p<.01), followed by compatibility with Current Medical Practice (r=.43, p<.01).

Linear associations were not observed between Professional Practice (private or group practice) and any of the other variables. Excluding this proposed moderator variable (Professional Practice), all of the predictor variables had a significant linear relationship with the criterion variable and with each other. Compatibility with Professional/Personal beliefs and Observability were the only other variables that were not related to each other (r=.01). The variables Relative Advantage of genomic medicine and compatibility with Current Medical Practice had moderate associations with Complexity (r=.33, p<.01 and r=.41, p<.01).

Compatibility with Professional/Personal beliefs had a small association with compatibility with Current Medical Practice ($r=.20$, $p<.01$). Complexity and Trialability of genomic medicine also had a small association with each other ($r=.28$, $p<.01$), as did Trialability and Observability of genomic medicine ($r=.14$, $p<.01$). Overall results of this test of linearity show only small to moderate correlations among the independent variables, which reduced potential problems of multicollinearity in the regression and path analysis.

Prediction of Likelihood of Primary Care Physicians' Adopting Genomic Medicine

The data were analyzed by multiple regression, using the dependent variable Likelihood of PCPs Adopting Genomic Medicine, all of the independent variables (Relative Advantage, Compatibility with Professional/Personal Beliefs and Values, Compatibility with Current Practice, Complexity, Trialability, and Observability), and the moderator variable Professional Practice (private or group practice) as predictors.

Table 31

Pearson Zero-Order Correlations Among Predictor and Criterion Variables

Variables	CV (Adopt)	P1	P2	P3	P4	P5	P6
P1 Relative Advantage	.337**						
P2 Compatibility with Prof/Pers beliefs	.170**	.261**					
P3 Compatibility with Current Med Practice	.434**	.295**	.205**				
P4 Complexity	.463**	.330**	.114*	.415**			
P5 Trialability	.270**	.156**	.136**	.361**	.287**		
P6 Observability	.282**	.170**	.011	.259**	.192**	.148**	
P7 Professional Practice	.060	.000	-.012	.002	.022	-.029	-.003

*p<.05

**p<.01

CV = Criterion Variable

P1-P7 = Predictor Variables

Table 32 presents a series of regression models estimating the net effects of demographic variables, perceived characteristics of genomic medicine and professional practice (private or group). In Model 1, Likelihood of Adopting Genomic Medicine is considered s a function of socio-demographic factors. In model 2, Relative Advantage was added as a predictor. Model 3 includes Compatibility with Professional/Personal beliefs and values and Compatibility with Current Practice as predictors. In Model 4 Complexity was added as a predictor, Trialability in Model 5, Observability in Model 6, and Professional Practice in Model 7.

None of the socio-demographic variables are significant predictors of Likelihood to Adopt Genomic Medicine. Four of the perceived characteristics of genomic medicine (Relative Advantage, Compatibility with Current Practice, Complexity, Observability) are associated with Likelihood to Adopt Genomic Medicine. For Models 4, 5, 6, and 7, Complexity was shown to be the strongest predictor of Likelihood to Adopt Genomic medicine. The effect of the Complexity variable is affected very little by the inclusion of the other variables in the model (Table 32).

Table 32

Metric and Standardized Beta Coefficients for Predictors of Likelihood of PCPs Adopting Genomic Medicine, According to Different Regression Models

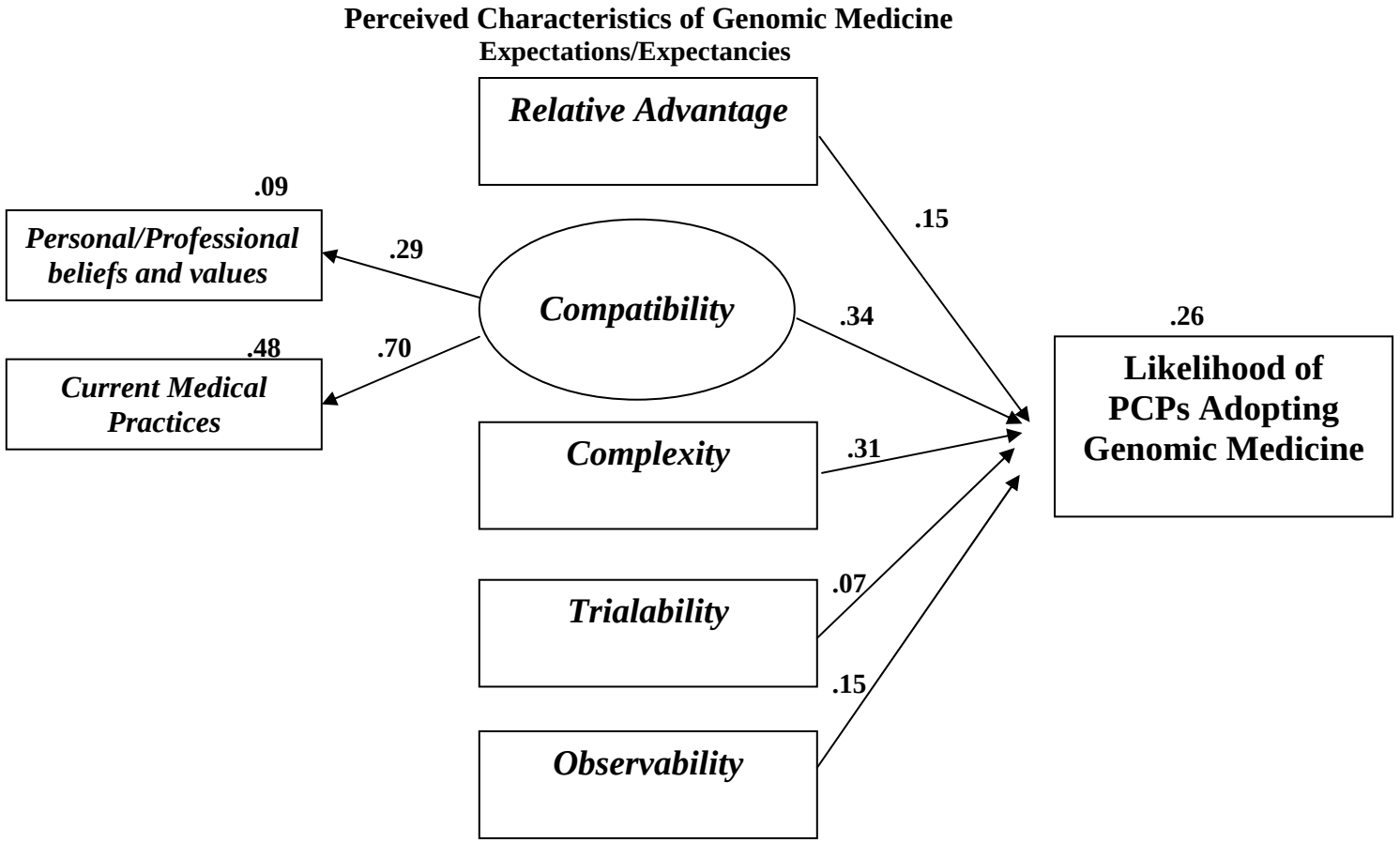
Predictors	*Model 1* *Adj R^2 = -.005*		*Model 2* *Adj R^2 = .104*		*Model 3* *Adj R^2= .224*		*Model 4* *Adj R^2= .295*		*Model 5* *Adj R^2= .299*		*Model 6* *Adj R^2= .314*		*Model 7* *Adj R^2=.317*	
	B	*β*	*B*	*β*	*B*	*β*	*B*	*β*	*B*	*β*	*B*	*β*	*B*	*β*
Constant	-5.815 (100.17)		18.330 (94.61)		-6.764 (88.19)		-17.144 (84.06)		-15.828 (83.86)		-6.093 (83.00)		-7.756 (82.82)	
Gender	-.419 (.432)	-.051	-.398 (.408)	-.049	-.387 (.380)	-.047	-198 (.363)	-.024	-.271 (.365)	-.037	-.195 (.362)	-.024	-228 (.362)	-.028
Age	-2.052E-03 (.051)	-.007	-9.582E-04 (.048)	- .003	7.407E-03 (.045)	.024	1.712E-02 (.043)	.054	1.472E-02 (.043)	.058	1.406E-02 (.043)	.045	1.760E-02 (.042)	.056
Graduation year	1.169E-02 (.049)	.038	-1.543E-03 (.047)	-.005	1.039E-02 (.043)	.034	1.507E-02 (.041)	.049	1.443E-02 (.041)	.049	9.075E-02 (.041)	.030	9.539E-02 (.041)	.031
Ethnicity	-8.014E-03 (.165)	-.002	1.046E-02 (.156)	.003	4.352E-02 (.146)	.014	-4.338E-02 (.139)	-.014	-6.446E-02 (.140)	-.016	-5.265E-02 (.138)	-.016	-3.907E-02 (.138)	-.012
Specialty	.146 (.119)	.062	8.974E-02 (.113)	.038	-3.057E-02 (.106)	-.013	-.104 (.102)	-.044	-.105 (.102)	-.045	-6.237E-02 (.102)	-.027	-6.218E-02 (.101)	-.026
Relative Advantage			.107 (.015)	.336***	7.029E-02 (.015)	.222***	4.810E-02 (.015)	.152**	4.783E-02 (.015)	.156**	4.411E-02 (.015)	.139**	4.568E-02 (.015)	.144**
Compatibility with Current Practice					8.082E-02 (.011)	363***	5.752E-02 (.011)	.258***	5.257E-02 (.011)	.233***	4.601E-02 (.011)	.207***	40530E-02 (.011)	.204***
Compatibility with Profess/ Personal beliefs					1.476E-02 (.019)	.035	1.815E-02 (.018)	.044	1.634E-02 (.018)	.037	2.078E-02 (.018)	.050	1.979E-02 (.018)	.048
Complexity							.110 (.017)	.308***	.105 (.017)	.289***	.101 (.017)	.284***	9.953E-02 (.017)	..279***
Trialability									3.980E-02 (.023)	.083	3.534E-02 (.023)	.070	3.735E-02 (023)	.074
Observability											3.973E-02 (.013)	.138**	3.937E-02 (.013)	.136**
Professional Practice													.362 (.219)	.070

* p<.05 ** p<.01 *** p<.001

Structural Equation Model

The Analysis of MOment Structures (AMOS 4.0) structural equation modeling software was also used for analysis and for testing the proposed model's fit to the data. Structural equation modeling (SEM) determines whether the model's implied covariance matrix of the measures is consistent with an empirical or data-based covariance matrix.[86] The technique also evaluates the contribution of each of the independent variables to the dependent variables.[85] Like multiple regression, the goal may be to predict which independent variable has the strongest influence on the dependent variable. Some of the variables can be latent, whereas others are directly observed. SEM assumes that all the relations are linear and that the underlying measurement and latent variables are continuous.[87]

The model for this study was comprised of Relative Advantage, Compatibility, a latent variable with 2 indicators (Compatibility with Professional/Personal Beliefs and Values and Compatibility with Current Practice), Complexity, Trailability, Observability, and Likelihood of PCPs Adopting Genomic Medicine (Figure 8). The model Chi Square value, also called *discrepancy*, represents the result of testing whether the implied variance structure is different from the observed (or measured) covariance matrix. Ideally, the two covariance matrices are the same (do not differ) and represent, therefore, a "good fit" for the model to the measured data. When interpreting results, therefore, researchers hope for a non-significant finding for the Chi-Square value. A statistically significant finding indicates that the two matrices/models differ substantially enough to indicate a "poor fit." In other words, a finding of significance means the



X^2 = 258.59, df = 14, p = 0.000, RMSEA = 0.64

Figure 8 Structural Equation Model for the Whole Sample

given model's covariance structure is significantly different from the observed covariance matrix.[88] The Chi Square for the model tested in this study showed that the overall fit of the proposed model was poor, $X^2 = 258.59$, df 14, $p = 0.000$. The root mean square error of approximation (RMSEA) is a measure of discrepancy per degree of freedom between the model and the population. A RMSEA of 0.05 or lower indicates a close fit.[88] The RMSEA of the proposed model was 0.64 further supporting the model's lack of fit. The model explained only 26% of the variance for likelihood of PCPs adopting genomic medicine. A nested model comparison for the two separate groups – private practice, group practice – was also calculated. The analysis revealed an equally poor fit for both groups.

Both the regression models and the SEM indicate that other factors must be measured when attempting to fully understand PCPs likelihood of adopting genomic medicine, such as insurance coverage, ethical, and political issues.

CHAPTER V

CONCLUSION, LIMITATIONS, AND DISCUSSION

Conclusion

It has been predicted that genomic medicine will change the future of health care. Achievements of the Human Genome Project are helping to identify the genes that contribute to common disorders such as diabetes, breast cancer, Alzheimer's, and cancer as well as genetic variants that influence a patient's response to a particular drug.[67,3,89] It has been projected that primary care physicians (PCPs) will play a larger role in predispositional diagnosis and management of many common disorders. These physicians will be using genetic testing routinely to determine the disorders their patients will someday develop.[90]

Clinical applications of genomic medicine will be adopted by PCPs at a variable pace. Characteristics of an innovation such as genomic medicine are strong indicators of its potential for adoption. Everett M. Roger's[11] Diffusion of Innovations theory defines an innovation as an idea, practice, or object that is perceived as new by members of a social system. According to Rogers, there are five characteristics of an innovation that influence the rate of its adoption; the *relative advantage* of the innovation, its *compatibility*, *complexity*, *trialability* and *observability*. The purpose of this study was to assess whether (and to what extent) physicians' perceptions of genomic medicine as an innovation influence their likelihood of adopting this innovation into primary care.

Findings from this study indicate that the Texas PCP who is most willing to adopt genomic medicine into his/her practice, strongly believes that genomic medicine

provides important advantages over traditional forms of medical practice, strongly perceives genomic medicine to be consistent with his/her professional and personal beliefs and values, and strongly perceives genomic medicine as a not-so-complex innovation. Additionally, the practitioner willing to adopt genomic medicine does not consider it very important to observe most of his or her colleagues adopting genomic medicine before considering adopting this innovation him/herself. These findings lend further support for the Diffusion of Innovations theory, which states that "innovators" require shorter adoption periods, have the ability to cope with a high degree of uncertainty about the innovation and the ability to understand and apply complex technical knowledge.[75] One of the responders commented on his/her survey: "I strongly believe genomic medicine will change the future of medicine. Treatment will change in the next five years from treating the patient after the disease to prevention of disease with genetic engineering."

The degree to which an innovation is perceived as better than what is presently being practiced can influence the rate of adoption.[11] According to this study, Texas primary care physicians believe one of the best advantages of genomic medicine is to supplement a family history. They also feel that taking a more detailed family history could easily be incorporated into their primary care practice. Khoury[91] maintains that a good "family history can build a bridge from genetics to genomics in practice."[(p265)] Previous family history can predict the risk factor for most chronic diseases such as coronary heart disease, diabetes, cancer, osteorporosis, and asthma. Yet family history is

rarely used in preventive medicine to assess disease risk or influence early detection and motivate prevention strategies.[91]

Wilkins-Haug and colleagues[92] found that gynecologists who take family histories to perform preconception screening also use that information to screen for increased risk of breast cancer, ovarian cancer, other cancers, and certain adult-onset disorders. However, only a small percentage of their respondents ordered DNA-based genetic tests to screen for disease even though they had patients who requested the tests.

In order for an innovation to be adopted it must be consistent with existing values, beliefs, and present practices of potential adopters.[11] Findings from this study indicate that offering termination of a pregnancy as an option even if the child would suffer from a severe mental or physical abnormality is not consistent with many Texas PCPs personal beliefs and values. Prenatal diagnosis and termination of pregnancy raises important ethical dilemmas when diagnosis and prognosis are uncertain. Deciding what is considered "severely abnormal" can cross that fine line to eugenics.

This study also found that although Texas PCPs perceive clinical applications of genomic medicine to be consistent with professional standards, components such as ordering genetic testing and genetic counseling are not compatible with their current practice. Genetic counseling is usually provided by physicians or counselors with advanced training in genetics.[93] Integrating this genetic service into practice requires additional time to interpret information about genetic disorders, analyze inheritance patterns and risks of recurrence, review available options with the patient's family and refer individuals and families to community or state-supported services.[94]

The anticipated explosion in genetic information will be a challenge for genetic counseling, no matter who provides it.[93] The more difficult it is to understand and use an innovation, the more reluctant potential adopters will be to embrace the change.[70] It was not a surprise that many Texas PCPs in this study find it difficult to stay updated on genomic-related knowledge. Many studies have identified inadequate knowledge of genetics, genetic testing, or genetic counseling as a barrier to incorporating genetic services in primary care.[34,35,95,96,97,98,99,100] In the study reported here, the obstetrics/gynecology and pediatric specialty group perceive genomic medicine to be less complex than the other specialty groups. The data suggest that the family medicine specialty group perceived genomic medicine to be more complex than the other specialty groups. Fetters and colleagues[101] found that many family medicine physicians feel there have been inadequate education opportunities to learn about genetics, genetic counseling, and the Human Genome Project. However, many of these physicians also indicated reluctance to invest in self-educational efforts until genetic problems become more relevant to their patients.

Texas PCPs in this study feel that genetic services could be incorporated into primary care practice on a gradual basis. Physicians are more likely to adopt an innovation if it is easy to try without having to fully commit to it or give up an existing practice. Considerable uncertainty exists about the purpose and value of genomic medicine as well as the evaluation and consequences of its use.[14] Trying out an innovation or new technology allows potential adopters to reduce their uncertainty about its risks and benefits.[14]

Applications of genomic medicine lack immediate visible results and are therefore not very observable. For example, a change in lifestyle may improve the long-term health outcome of a patient found to be susceptible to Type II diabetes but show no immediate changes in health. "The willingness of physicians to offer an accurate, low-cost test to predict future disease, even if no others in their specialty do, has implications for the diffusion of new genetic technology, at least in the initial stages."[102 (p1000)] With the exception of the obstetric/gynecology specialty, most of the respondents in the present study were not currently observing colleagues adopting genetic services or assisting patients to make decisions regarding genetic services.

Likelihood of PCPs Adopting Genomic Medicine

Texas primary care physicians in this study were likely to order carrier testing (55.5%), to order predictive testing (64.5%), and to refer patients for a genetic consultation (80.5%). Almost half (49%) were likely to order preconception counseling. The present study found that the obstetrics/gynecology specialty group was more likely than the other specialty groups to adopt all clinical uses of genomic medicine. Although PCPs in all specialty groups (internal medicine, pediatrics, obstetrics/gynecology, family medicine, general practice, other specialty) felt an advantage of genomic medicine is to diagnose a genetic condition in an embryo before it is implanted, only 20.5% (mostly obstetrics and gynecology PCPs) were likely to order preimplantion diagnosis for their patients.

Preimplantation genetic diagnosis (PGD) is offered as an alternative to prenatal diagnosis to avoid the risk for pregnancy termination.[103] PGD is presently applicable

for numerous genetic-related disorders. For example, PGD has been performed for couples carrying gene mutations known to determine a strong predisposition to most cancers. Applications of this genetic technology has also been indicated for neurofibromatosis, a common autosomal-dominant neurological disorder, as well as early-onset Alzheimer's disease, an autosomal dominant familial predisposition to presenile dementia.[103]

A PGD-assisted pregnancy can ensure that the resulting baby is human leukocyte antigen (HLA)-matched to an affected sibling in need of a bone marrow transplantation.[103] However, both stem cell research and preimplantation diagnosis are entangled in the debate over abortion, and the general question of when human life begins. Another controversial use of preimplantation diagnosis involves identifying and choosing gender for non-therapeutic purposes. This practice is considered discriminatory and not in the best interest of society nor for the resulting child.[45]

Limitations

While this study examined a randomly selected sample of primary care physicians in the state of Texas, one of its limitations is the possibility that physicians responding to the survey had more genetics knowledge or stronger interest in genomic medicine than the nonresponders. Because of the significant difference between the ethnicities of the responders and nonresponders, this study is also limited in its capability to generalize across all ethnicities of primary care physicians. A comparison of the total random sample (1371) showed Black responders to be significantly underrepresented (5.0%) when compared to White (33.8%), Hispanic (22%), and Asian (24%) responders.

The model proposed for this study only accounted for 26% of the variance in the structural equation analysis of likelihood of primary care physicians adopting genomic medicine (and 32% in the multiple regression model containing all proposed variables.) Since it was necessary to keep the survey brief, this study only examined the perceived characteristics of the innovation of genomic medicine. Rates of diffusion and adoption depend to a large extent on how certain characteristics of the innovation interact with the targeted social system. Because the behaviors or characteristics of the system's individual members (i.e., perceived norms, personal innovativeness) also are crucial for understanding the adoption process,[70] future research is needed to examine the characteristics of the "innovators," which may account for a larger portion of the variance in the likelihood of adoption of genomic medicine. Previous studies found that "early adopters" or "innovators" are able to cope with a high degree of uncertainty about an innovative technology such as genomic medicine.[102]

An interesting finding in this study was that Texas PCPs are similar in their perceptions about genomic medicine regardless of their practice setting (group, private.) However, the choice of practice setting (private or group) as a possible moderator variable proved to be a weakness for the study's proposed model. Data analysis showed that the social system that actually had the most effect on the other variables was the specialty group (internal medicine, pediatric, obstetrics/gynecology, family medicine, general practice, or other specialty.) However, use of "specialty group" as a moderator variable to examine nested models was not included in the structural equation analysis due to the insufficient numbers in each group's sample. Other characteristics of the primary care physician's social network (e.g., managed care organizations, government institutions) as well as their communication channels (journals, professional societies and conferences) also need to be considered in the adoption and diffusion of genomic medicine.

Moreover, the ethical, legal, and social issues of genomic medicine as well as the funding of genetic services were not explored in this study. However, comments from several respondents made it clear that these characteristics of genomic medicine cannot be overlooked in the adoption and diffusion process:

> HMO/PPO either won't pay [for genetic services] or for sure do not pay for the time it takes to do this. The real world of medicine isn't very good!
>
> Providing genetic services is a time consuming practice with low reimbursements. Also border town issues such as no insurance coverage.
>
> [Genetic services are] usually driven by insurance provider contracts.
>
> If I get reimbursed by insurance companies for the time I will spend doing all these [genetic services], I will be happy to do so.

Discussion

The transition from traditional medicine to genomic medicine will require most PCPs to understand the language of the "new genetics"[95] and receive ongoing support to communicate effectively with patients and their families. Mann[104] calls for a new model of health care to be developed incorporating PCPs, specialists, and allied health care professionals in order to provide relevant genetic information and procedures to the patients and community. As primary care becomes more focused on prevention, the future of health care also will need to include the health promotion specialist. For Guttmacher,[105] "Health educators will play a key role in shaping messages about individualized health maintenance that will be critical to the full flowering of genomic health care." (p220)

Nevertheless the emphasis on concepts of risk and disease predisposition will be a challenge for health educators. While clinical genetic specialists are experienced in patient education, health educators have more experience in affecting health behaviors.[106] Much of the power of genomic medicine will rely on the ability to change behaviors (including lifestyle and diet) based upon predisposition for health risks.[105] Caumartin and colleagues[107] call for students of all disciplines to add to their discipline-specific skills an understanding of the role genetics will play. The authors add that for health behavior/health education specialists, genetic roles may include:[107]

- Assessing public demand for genetic information.
- Educating the public on genetic issues.
- Evaluating the psychosocial impact of this information on individuals, families, and populations.
- Developing educational strategies that will communicate the complexities of genetics to the lay public.

- Developing effective behavior modifications that prevent disease in those with an inherited susceptibility. The goal will be to assist the public in making reasoned decisions about their use of genetic testing, understanding test results, and analyzing the impact that test results may have on their lives.
- Contributing to an understanding of individual variation in the utilization of genetic services.
- Taking a key role in defining the ethical and social dimensions and implications of genetic technology.

The impact of genetics on health care delivery and training, and on the role of primary care physicians provides a major challenge for those charged with promoting new genetic technologies. There is a legitimate need to control genetic technology diffusion while research is conducted to assess clinical utility and cost effectiveness (versus pressures for adoption created by the media), public demand, producers and the profession.[108]

"It is clear that both the public and health professionals dealing with the public need further education regarding the role of genetics in health and disease."[106 (p90)] As Guttmacher[105] highlights, however, "the expansion of nongenetic specialist providers' use of genetics will not relegate genetic specialists to the dustbin of medical history, but instead will redefine their roles."[(p218)] The diagnosis and long-term follow-up of

individuals with monogenic and chromosomal conditions will continue to require the specialized knowledge that remains part of the genetic specialist's practice.[105]

As genetic information becomes more relevant to common disorders, PCPs will perceive an expanded role for themselves in genomic medicine such as evaluating genetic risk, counseling patients about testing, ordering tests, and referring patients for genetic consultation.[109] The present study indicates that greater emphasis will need to be placed on knowledge pertinent to genomic medicine in medical education curricula. It will also be important for continuing education programs to be developed and offered to primary care physicians to keep them updated on new advances in genomic medicine as well as information regarding genetic services in their area. Burke and Emery[43] assert that relevant genetics education for primary care would include information about the indicators of genetic disease and the rationale for including genetic disorders in the differential diagnosis of common problems. For the already time-constrained physician, sources for quick reference when a patient presents with an unusual clinical problem or family history would also be helpful.

Test Results, Positive

There is no magic in my bag
No aces up these white coat sleeves
No healing spells, no tricks to please
No Merlin's song to save the day.

To comfort you should be my trick
Should be our goal, our common boast.
To do no harm, my solemn oath
Is jeopardized by what I know.

The words I chant will shake your soul
Will bubble forth to change your life
Like sorcerers with beards of white
Will make you yearn for days gone by.

The news I bring is from the void.
It summons grief, directs the storms.
A crimson cape of life's dreams torn
This wizard waves before your eyes.

I mix my brew, you toss it down.
The genie's out, the truth is loose,
Your perfect health: a painful ruse.
No magic words will save you now.

By J. Trig Brown, M.D.[110]

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APPENDIX A

GLOSSARY OF GENETIC TERMS

GLOSSARY OF GENETIC TERMS

Alzheimer's Disease

This is the most common cause of dementia in those over 65. The condition is more common in women. The disease usually presents with memory loss and a gradual progressive deterioration in functioning

Amniocentesis

Prenatal diagnosis method using cells in the amniotic fluid to determine the number and kind of chromosomes of the fetus and, when indicated, perform biochemical studies.

Autosomal Recessive Gene

A gene which will be expressed only if there are 2 identical copies or, for a male, if one copy is present on the X chromosome.

BRCA1/BRCA2

The first breast cancer genes to be identified. Mutated forms of these genes are believed to be responsible for about half the cases of inherited breast cancer, especially those that occur in younger women. Both are tumor suppressor genes.

Carrier -- an individual heterozygous for a single recessive gene.

Carrier Testing

Carrier testing is performed to identify individuals who have a gene mutation for a disorder inherited in an autosomal recessive or X-linked recessive manner. Carriers usually do not themselves have symptoms related to the gene mutation. Carrier testing is offered to individuals who have family members with a genetic condition, family members of an identified carrier, and individuals in ethnic or racial groups known to have a higher carrier rate for a particular condition.

Chromosome -- in the eukaryotic nucleus, one of the threadlike structures consisting of chromatin and carry genetic information arranged in a linear sequence.

Chorionic Villus Sampling

An invasive prenatal diagnostic procedure involving removal of villi from the human chorion to obtain chromosomes and cell products for diagnosis of disorders in the human embryo.

Cystic Fibrosis

An autosomal recessive genetic condition of the exocrine glands, which causes the body to produce excessively thick, sticky mucus that clogs the lungs and pancreas, interfering with breathing and digestion.

Disease -- any deviation from the normal structure or function of any part, organ, or system of the body that is manifested by a characteristic set of symptoms and signs whose pathology and prognosis may be known or unknown.

Diabetes Millenitis

Two types of a highly variable disorder in which abnormalities in the ability to make and/or use the hormone insulin interfere with the process of turning dietary carbohydrates into glucose, the body's fuel. Type I is known as insulin dependent diabetes mellitus, and type II is known as non-insulin dependent diabetes mellitus.

Diagnostic Testing

Diagnostic testing is used to confirm or rule out a known or suspected genetic disorder in a symptomatic individual.

Double Helix

The structural arrangement of DNA, which looks something like an immensely long ladder twisted into a helix, or coil. The sides of the "ladder" are formed by a backbone of sugar and phosphate molecules, and the "rungs" consist of nucleotide bases joined weakly in the middle by hydrogen bonds.

DNA Sequencing

"Plus and minus" or "primed synthesis" method, developed by Sanger, DNA is synthesized in vitro in such a way that it is radioactively labeled and the reaction terminates specifically at the position corresponding to a given base; the "chemical" method, ssDNA is subjected to several chemical cleavage protocols that selectively make breaks on one side of a particular base.

Down Syndrome

A type of mental deficiency due to trisomy (three copies) of autosome 21, a translocation of 21 or mosaicism.

Duchenne/Becker Muscular Dystrophy

The most common and severe form of muscular dystrophy; transmitted as an X-linked trait. X-linked recessive. Symptoms include onset at 2-5 years with difficulty with gait and stairs, enlarged calf muscles, progression to wheelchair by adolescence, shortened life span.

ELSI

Ethical, legal and social implications (of HGP).

Ethics

The study of fundamental principles which defines values and determines moral duty and obligation.

Eugenics

The improvement of humanity by altering its genetic composition by encouraging breeding of those presumed to have desirable genes.

Fragile-X Syndrome

X-linked trait; the second most common identifiable cause of genetic mental deficiency.

Gene

A hereditary unit that occupies a certain position on a chromosome; a unit that has one or more specific effects on the phenotype, and can mutate to various allelic forms.

Gene Therapy

Addition of a functional gene or group of genes to a cell by gene insertion to correct an hereditary disease.

Genetic Counseling

The educational process that helps individuals, couples, or families to understand genetic information and issues that may have an impact on them.

Genetic Screening

Testing groups of individuals to identify defective genes capable of causing hereditary conditions.

Genetic Variation

A phenotypic variance of a trait in a population attributed to genetic heterogeneity.

Genetics

The study of inheritance patterns of specific traits.

Genome

All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

Genomics

The study of genes and their function.

Hemochromatosis

The most common form of iron overload disease, is an inherited disorder that causes the body to absorb and store too much iron. The extra iron builds up in organs and damages them. Without treatment, the disease can cause these organs to fail.

Hemoglobin Disorder

Hemoglobin is a respiratory protein contained in red blood cells that transports oxygen from the lungs to the tissues of the body. A Hemoglobin disorder is a condition caused by a defect in the genetic code for hemoglobin synthesis. This defect affects the amount or the quality of the hemoglobin being produced. Sickle cell anemia is a common hemoglobin disorder. Other common hemoglobin disorders include hemoglobin SC disease and S-beta thalassemia.

Hemophilia

The hemophilias are inherited disorders that cause abnormal bleeding. Symptoms range from increased bleeding after trauma, injury, or surgery to sudden bleeding with no apparent cause. The two types of hemophilia are hemophilia A (also called classic hemophilia) and hemophilia B (also called Christmas disease). Hemophilia A is more common -- about 85% of people who have hemophilia have this form.

Human Leukocyte Antigen (HLA)

Proteins located on the surface of white blood cells which play an important role in our body's immune response to foreign substances. These antigens are also used to determine the suitability of a match between an donor and a recipient. Patients and potential donors have their white blood cells tested for three antigens - HLA-A, -B and -DR. Each individual has two sets of these antigens, one set inherited from each parent. For this reason, it is much more likely for a brother or sister to match the patient than an

unrelated individual, and much more likely for persons of the same racial and ethnic backgrounds to match each other.

Human Genome Project

The joint national effort, led by DOE and NIH, begun in 1986 by DOE to create an ordered set of DNA segments from known chromosomal locations, develop new computational methods for analyzing genetic map and DNA sequence data, and develop new techniques and instruments for detecting and analyzing DNA.

Huntington Disease

A disease characterized by irregular, spasmodic involuntary movements of the limbs and facial muscles, mental deterioration and death, usually within 20 years of the onset of symptoms.

Marfan Syndrome -- autosomal dominant condition of connective tissue; affects the skeletal, ocular and cardiovascular systems.

Mendelian Inheritance

One method in which genetic traits are passed from parents to offspring. Named for Gregor Mendel, who first studied and recognized the existence of genes and this method of inheritance.

Monogenic

A disorder caused by mutation of a single gene.

Multifactorial

A characteristic influenced in its expression by many factors, both genetic and environmental.

Mutation

Process by which genes undergo a structural change.

Neurofibromatosis

One of the most common single gene conditions affecting the human nervous system; in most cases, "cafe au lait" spots, are the only symptom; inherited as an autosomal dominant trait, with 50% being new mutations.

Newborn Screening

Newborn screening identifies individuals who have an increased chance of having a specific genetic disorder so that treatment can be started as soon as possible.

Points to consider:

- ◊ Newborn screening programs are usually legally mandated and vary from state to state.
- ◊ Newborn screening is performed routinely at birth, unless specifically refused by the parents in writing.
- ◊ Screening tests are not designed to be diagnostic, but to identify individuals who may be candidates for further diagnostic tests.
- ◊ Many parents do not realize that newborn screening has been done (or which tests were included), even if they signed a consent form when their child was born.

- ◊ Education is necessary with positive screening results in order to avoid misunderstandings, anxiety and discrimination.

Pharmacogenomics

The study of the interaction of an individual's genetic makeup and response to a drug.

Polygenic Disorder

Genetic disorder resulting from the combined action of alleles of more than one gene (e.g., heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns usually are more complex than those of single-gene disorders.

Predictive Testing

Predictive testing is offered to asymptomatic individuals with a family history of a genetic disorder. Predictive testing is of two types: presymptomatic (eventual development of symptoms is certain when the gene mutation is present, e.g., Huntington disease) and predispositional (eventual development of symptoms is likely but not certain when the gene mutation is present, e.g., breast cancer).

Predisposition -- to have a tendency or inclination towards something in advance.

Preimplantation Testing (Preimplantation Genetic Diagnosis)

Preimplantation testing is performed on early embryos resulting from in vitro fertilization in order to decrease the chance of a particular genetic condition occurring in the fetus. It is generally offered to couples with a high chance of having a child with a serious disorder. Preimplantation testing provides an alternative to prenatal diagnosis and termination of affected pregnancies.

Prenatal Testing

Prenatal testing is performed during a pregnancy to assess the health status of a fetus. Prenatal diagnostic tests are offered when there is an increased risk of having a child with a genetic condition due to maternal age, family history, ethnicity, or a suggestive fetal ultrasound examination. Routine prenatal diagnostic test procedures are amniocentesis and chorionic villus sampling (CVS).

Presymptomatic Diagnosis

Diagnosis of a genetic condition before the appearance of symptoms.

Retinoblastoma

Occurs in early childhood and affects about 1 child in 20,000. The tumor develops from the immature retina - the part of the eye responsible for detecting light and color. There are both hereditary and non-hereditary forms of retinoblastoma. IN the hereditary form,

multiple tumors are found in both eyes, while in the non-hereditary form only one eye is effected and by only one tumor.

Sickle Cell Anemia

An hereditary, chronic form of hemolytic anemia characterized by breakdown of the red blood cells; red blood cells undergo a reversible alteration in shape when the oxygen tension of the plasma falls slightly and a sickle-like shape forms.

Single-Gene Disorder

Hereditary disorder caused by a mutant allele of a single gene (e.g., Duchenne muscular dystrophy, retinoblastoma, sickle cell disease).

Stem cell

Undifferentiated, primitive cells in the bone marrow that have the ability both to multiply and to differentiate into specific blood cells.

Syndrome

A recognizable pattern or group of multiple signs, symptoms or malformations that characterize a particular condition; syndromes are thought to arise from a common origin and result from more than one developmental error during fetal growth.

Tay-Sachs disease

A fatal degenerative disease of the nervous system due to a deficiency of hexosamidase A, causing mental deficiency, paralysis, mental deterioration, and blindness; found primarily but not exclusively among Ashkenazi Jews. Autosomal recessive.

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Debra Collins, M.S. CGC, Genetic Counselor, dcollins@kumc.edu
http://www.kumc.edu/gec/glossnew.html

National Human Genome Research Institute
http://www.genome.gov/glossary.cfm
Human Genome Project Information
http://www.ornl.gov/TechResources/Human_Genome/glossary/

GeneTests
Funded by the National Institutes of Health, the Human Resource and Service Administration, and the Department of Energy.
http://geneclinics.org

Psychejam.Com
http://www.psychejam.com/altzheimer's.htm

Glossary: Health on the Net Foundation
http://www.hon.ch/Library/Theme/Allergy/Glossary/hla.html

Primary Care and Family Health
GENETIC DISEASE BRANCH
Glossary of Selected Medical Terms
http://www.dhs.cahwnet.gov/pcfh/gdb/html/GDB/screening.htm

National Digestive Diseases Information Clearinghouse (NDDIC)
A service of the National Institute of Diabetes and Kidney Diseases (NIDDK)
http://digestive.niddk.nih.gov/ddiseases/pubs/hemochromatosis/index.htm

WebbMD
http://aolsvc.health.webmd.aol.com/content/article/7/1680_53571.htm?SRC=aolKW=hemophilia

Genes and Disease
http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View.ShowSection&rid=gnd.section.129

APPENDIX B

TEXAS A&M INSTITUTIONAL REVIEW BOARD

VPR
Office of the Vice President for Research

February 19, 2003

MEMORANDUM

TO: Sandra Suther
HLKN
MS 4243

SUBJECT: Genomic Medicine in Primary Care: Texas Physicians' Adoption of an Innovation
2003-0084

Approval Date: February 19, 2003 to February 18, 2004

The Institutional Review Board – Human Subjects in Research, Texas A&M University has reviewed and approved the above referenced protocol. Your study has been approved for one year. As the principal investigator of this study, you assume the following responsibilities:

Renewal: Your protocol must be re-approved each year in order to continue the research. You must also complete the proper renewal forms in order to continue the study after the initial approval period.

Adverse events: Any adverse events or reactions must be reported to the IRB immediately.

Amendments: Any changes to the protocol, such as procedures, consent/assent forms, addition of subjects, or study design must be reported to and approved by the IRB.

Informed Consent/Assent: All subjects should be given a copy of the consent document approved by the IRB for use in your study.

Completion: When the study is complete, you must notify the IRB office and complete the required forms.

Dr. E. Murl Bailey, CIP, Advisor
Institutional Review Board –
Human Subjects in Research

APPENDIX C

INTERVIEW GUIDES FOR QUALITATIVE STUDY

Interview Guide for Physicians, Nurses and Social Workers
In-Depth Interviews
Qualitative Phase

Beliefs and Training Needs of Health Professionals
Regarding Genetics and Public Health:
Developing and Validating a Survey Instrument

Question Type	Wording
Opening statement	The objective of this study is to learn about health professionals' beliefs, knowledge, training needs and perceptions of their roles concerning the relationship between new genetic developments (the "new genetics") and public health. We are also interested in learning how providers perceive the potential impact of the "new genetics" on their daily practice, and their views on prevention of genetic-related health problems.
Opening question	Please tell us your name and a little bit about what you do and where you do it. Probe: name of institution/place of employment Specialization Practice How long in current practice?
	Impact on Public Health
Introductory	Given the recent discoveries and developments in the study of human genetics, how do you see these developments affecting public health?
Key	Do you have any concerns regarding this impact of genetic discoveries on public health? Probe: Any concerns that media attention to genetic issues may raise public anxiety and increase the demand for genetic services?
	Impact on Provider's Own Practice
Key	How do you see this "new genetics" (as these developments have been recently termed) affecting what you do here on a regular basis? Probe: Workload implications Training
Key	What are some of the barriers that professionals like you may encounter when dealing with genetic-related issues (either prevention or treatment/management)?
Key	When you see a client who you suspect may need to deal with genetic issues, what do you do? Probe: Counsel Refer

Question Type	Wording
Key	IF REFER: Who would/do you refer this person to? Probe: Genetic Services Specialists in the field of concern (e.g., ophthalmology; oncology) Colleagues for second opinion
Key	As far as you know, are there any genetic services available to you for consult and/or for referral of patients?
	Training Needs
Transition	This "new genetics" is producing quite a bit of new information. How do you see yourself handling this new information? Probe: CEU Additional Training Referring/Not dealing with it
Key	Have you attended any continuing education courses in genetics? Probe: CEU Conference presentations
Key	Do you feel you need additional training in genetics? If so – in which aspects would they need additional training?
Key	For you, what would be the best way to receive additional training? Probe: CEUs Teleconferences Professional Conferences Workshops In-services Academic Detailing
	Prevention
Transition	When I say "prevention of genetic-related health problems", what comes to mind?
Key	As you see it, who will have the responsibility to focus on prevention of genetic-related health problems? Probe: Do they perceive their role as preventive or curative/management? Is prevention the job of the health educator / nurse? Should genetic testing be patient- or physician-driven?
Key	In your opinion, what are the most effective strategies for prevention of genetic-related health problems?
Key	What is the role of family planning, in the prevention of genetic-related health problems?
	Un-Addressed Concerns
Ending	Are there any concerns or other issues that are important for us to address in a survey of health care providers, concerning this topic?

Interview Guide for Geneticists
In-Depth Interviews
Qualitative Phase

Beliefs and Training Needs of Health Professionals Regarding Genetics and Public Health: Developing and Validating a Survey Instrument

Question Type	Wording
Opening statement	The objective of this study is to learn about health professionals' beliefs, knowledge, training needs and perceptions of their roles concerning the relationship between new genetic developments (the "new genetics") and public health. We are also interested in learning how providers perceive the potential impact of the "new genetics" on their daily practice, and their views on prevention of genetic-related health problems.
Opening question	Please tell us your name and a little bit about what you do and where you do it. Probe: name of institution/place of employment Specialization Practice How long in current practice?
	Impact on Public Health
Introductory	Given the recent discoveries and developments in the study of human genetics, how do you see these developments affecting public health?
Key	Do you have any concerns regarding this impact of genetic discoveries on public health? Probe: Any concerns that media attention to genetic issues may raise public anxiety and increase the demand for genetic services?
	Impact on Provider's Own Practice
Key	How do you see this "new genetics" (as these developments have been recently termed) affecting what primary care providers do in their practice? Workload implications Training
Key	What are some of the barriers that primary care professionals may encounter when dealing with genetic-related issues (either prevention or treatment/management)?
	Training Needs
Key	Do you feel PCPs need additional training in genetics? If so – in which aspects would they need additional training?

Question Type	Wording
Key	What would be the best way for PCPs to receive additional training? Probe: CEUs Teleconferences Professional Conferences Workshops In-services Academic Detailing
	Prevention
Transition	When I say "prevention of genetic-related health problems", what comes to mind?
Key	As you see it, who will have the responsibility to focus on prevention of genetic-related health problems? Probe: Do they perceive their role as preventive or curative/management? Is prevention the job of the health educator / nurse? Should genetic testing be patient- or physician-driven?
Key	In your opinion, what are the most effective strategies for prevention of genetic-related health problems?
Key	What is the role of family planning, in the prevention of genetic-related health problems?
	Un-Addressed Concerns
Ending	Are there any concerns or other issues that are important for us to address in a survey of health care providers, concerning this topic?

Interview Guide for Health Educators
In-Depth Interviews
Qualitative Phase

Beliefs and Training Needs of Health Professionals Regarding Genetics and Public Health: Developing and Validating a Survey Instrument

Question Type	Wording
Opening statement	The objective of this study is to learn about health professionals' beliefs, knowledge, training needs and perceptions of their roles concerning the relationship between new genetic developments (the "new genetics") and public health. We are also interested in learning how providers perceive the potential impact of the "new genetics" on their daily practice, and their views on prevention of genetic-related health problems.
Opening question	Please tell us your name and a little bit about what you do and where you do it. Probe: name of institution/place of employment Area of Specialization How long in current position?
	Impact on Public Health
Introductory	Given the recent discoveries and developments in the study of human genetics, with the discovery of several disease-causing genes for instance, how do you see these developments affecting public health?
Key	Do you have any concerns regarding this impact of genetic discoveries on public health?
Key	How do you see this "new genetics" (as these developments have been recently termed) affecting your work as a health educator?
	How do you see the role of health education in the future, considering these new genetic developments we're seeing?
Key	What are some of the barriers that professionals like you may encounter when dealing with genetic-related issues (either prevention or treatment/management)?
	Training Needs
Transition	This "new genetics" is producing quite a bit of new information. How do you see yourself handling this new information? Probe: CEU Additional Training Referring/Not dealing with it

Question Type	Wording
Key	Have you attended any continuing education courses on genetics? Probe: CEU Conference presentations
Key	Do you feel you need additional training in genetics? If so – in which aspects would you like to have additional training?
Key	What would be the best way to receive additional training? Probe: CEUs Teleconferences Professional Conferences Workshops In-services Academic Detailing
	Prevention
Transition	When I say "prevention of genetic-related health problems", what comes to mind?
Key	As you see it, who will have the responsibility to focus on prevention of genetic-related health problems?
Key	In your opinion, what are the most effective strategies for prevention of genetic-related health problems?
Key	What is the role of family planning, in the prevention of genetic-related health problems?
	Un-Addressed Concerns
Ending	Are there any concerns or other issues that are important for us to address in a survey of health educators, concerning this topic?

APPENDIX D

INSTRUMENT

Genomic Medicine in Primary Care

This section is designed to assess your beliefs about the advantages of genomic medicine.

(Circle the number that best reflects your answer)

1. I believe one of the advantages of genomic medicine is to...	**Strongly Agree**	**Agree**	**Disagree**	**Strongly Disagree**	**I'm Not Sure**
a. Diagnose a genetic condition in an embryo before it is implanted instead of waiting and doing an ultrasound later in the pregnancy.	1	2	3	4	5
b. Perform carrier testing for a possible autosomal recessive disorder before the onset of symptoms.	1	2	3	4	5
c. Supplement a family history in predicting the risk of a healthy individual developing a disease.	1	2	3	4	5
d. Supplement knowledge of previous medical history in predicting which medications will be most effective for specific patients.	1	2	3	4	5

2. How important is it for you to be able to...	**Extremely Important**	**Somewhat Important**	**Not Very Important**	**Not Important At All**	**I'm Not Sure**
a. Detect a genetic condition in an embryo before it is implanted?	1	2	3	4	5
b. Test whether an individual possesses a copy of a mutated gene for an autosomal recessive disorder before the onset of symptoms?	1	2	3	4	5
c. Supplement a family history in predicting the risk of a healthy individual developing a disease?	1	2	3	4	5
d. Supplement previous knowledge of medical history in predicting which medications will be most effective for specific patients?	1	2	3	4	5

Compatibility of genomic medicine	**Strongly Agree**	**Agree**	**Disagree**	**Strongly Disagree**	**I'm Not Sure**
3. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is consistent with my professional standards.	1	2	3	4	5
4. Offering predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is consistent with my professional standards.	1	2	3	4	5

(Circle the number that best reflects your answer)	Strongly Agree	Agree	Disagree	Strongly Disagree	I'm Not Sure
5. Termination of pregnancy when there is a substantial risk that if a child were born it would suffer from a serious mental or physical abnormality is compatible with my personal values.	1	2	3	4	5
6. Predictive testing for diseases in which there is no available treatment or cure (such as Huntington's Disease) is compatible with my personal values.	1	2	3	4	5

	Extremely Important	Somewhat Important	Not Very Important	Not Important At All	I"m Not Sure
7. How important is it to you for genomic medicine to be consistent with your professional standards?	1	2	3	4	5
8. How important is it to you for genomic medicine to be compatible with your personal values?	1	2	3	4	5
9. How important is it to you for genomic medicine to be easily incorporated into your primary care practice?	1	2	3	4	5

	Strongly Agree	Agree	Disagree	Strongly Disagree	Already Incorporated
10. Genetic counseling could easily be incorporated into my primary care practice.	1	2	3	4	5
11. Taking a more detailed family history could easily be incorporated into my primary care practice.	1	2	3	4	5
12. Genetic testing could easily be incorporated into my primary care practice.	1	2	3	4	5

Complexity of genomic medicine

13. How easy or difficult is it for you to...	Extremely Easy	Somewhat Easy	Somewhat Difficult	Extremely Difficult	I'm Not Sure
a. Locate available genetic services?	1	2	3	4	5
b. Stay updated on genomic medicine-related knowledge?	1	2	3	4	5

	(Circle the number that best reflects your answer)				
14. How important is it for you...	**Extremely Important**	**Somewhat Important**	**Not Very Important**	**Not Important At All**	**I"m Not Sure**
a. To be able to locate available genetic services without difficulty?	1	2	3	4	5
b. To easily stay updated on genomic medicine-related knowledge?	1	2	3	4	5

Trialability of genomic medicine	**Strongly Agree**	**Agree**	**Disagree**	**Strongly Disagree**	**I'm Not Sure**
15. Genetic services can gradually be incorporated into primary care practice.	1	2	3	4	5
16. Genetic technologies, unlike other medical technologies, cannot be incorporated on a trial basis.	1	2	3	4	5
17. How important is it for you to...	**Extremely Important**	**Somewhat Important**	**Not Very Important**	**Not Important At All**	**I'm Not Sure**
a. Be able to gradually incorporate genetic services into your practice.	1	2	3	4	5
b. Incorporate technologies that you have tried first?	1	2	3	4	5

Observability of genomic medicine	**Strongly Agree**	**Agree**	**Disagree**	**Strongly Disagree**	**I'm Not Sure**
18. Most of my colleagues are...					
a. Adopting genetic testing into their practice.	1	2	3	4	5
b. Assisting patients to make decisions regarding genetic services.	1	2	3	4	5
19. Before you consider adopting genomic medicine into your practice, how important is it for you that your colleagues...	**Extremely Important**	**Somewhat Important**	**Not Very Important**	**Not Important At All**	**I'm Not Sure**
a. Adopt genetic testing into their practice?	1	2	3	4	5
b. Assist patients in making decisions regarding genetic services?	1	2	3	4	5

(Circle the number that best reflects your answer) **20. For your patients, how likely are you to...**	Extremely Likely	Somewhat Likely	Not Likely	Not Likely At All	I Already Am
a. Order carrier testing for a possible autosomal recessive disorder?	1	2	3	4	5
b. Order a preimplantation diagnosis to check for genetic disease in an embryo?	1	2	3	4	5
c. Order a predictive test for risk of disease?	1	2	3	4	5
d. Provide pre-conception counseling?	1	2	3	4	5
e. Refer them for a genetic consultation?	1	2	3	4	5

21. Which best describes your professional practice?

____ Private practice
____ Group practice
____ Other _____

Tell us about yourself...

22. Are you male or female?

____ Male
____ Female

23. What is your age? _____

24. What year did you graduate from medical school?

_____

25. Which specialty best describes you?

___ Internal medicine
___ Pediatrics
___ Obstetrics
___ Gynecology
___ Obstetrics/Gynecology
___ Family Medicine
___ General Practice
___ Other _____

26. Texas County in which you practice _____

APPENDIX E

COVER LETTER AND INFORMED CONSENT FORM

FOR PILOT AND FINAL STUDY

TEXAS A&M UNIVERSITY
College of Education and Human Development
Department of Health & Kinesiology

[Date]

Dear Dr.

I need your help with my dissertation research!

If you would take a few minutes to fill out this **short but very important** survey, it would be very appreciated. Results of the survey will be used to examine the factors that influence the likelihood of physicians incorporating genomic medicine into their primary care practice. This survey does **not** intend to measure knowledge about genetic medicine.

You are being invited to participate in this survey (along with 50 peers) because your name was recommended by an acquaintance. All surveys will be number coded but your answers are completely confidential. Only statistical information from aggregated data will be used for reporting.

If you have any questions about the survey, feel free to call me or write me at the address below. If you prefer, the completed survey can be faxed back.

Thank you very much for helping with this important study.

Sincerely,

Sandra Suther, M.A.
Texas A&M University
Department of Health & Kinesiology
4243 Tamu
College Station, TX 77843-4243
Phone: 210-573-0618
Fax: 979-696-0618
ssuther@tamu.edu

TEXAS A&M UNIVERSITY
College of Education and Human Development
Department of Health & Kinesiology

Informed Consent for Pilot Study Survey Participants
Genomic Medicine in Primary Care: Texas Physicians' Adoption of an Innovation

DATE, 2003

I have been invited to participate in a pilot study of the views that primary care physicians in Texas have of genomic medicine.

Sandra Suther is conducting this study. Presently, she is a doctoral student in health education and health promotion in the Department of Health and Kinesiology, College of Education and Human Development, at Texas A&M University. One of her main research interests is public health genetics. This study is part of her dissertation research.

The recent completion of the first draft of the human genome sequence is generating concerns among health communities world-wide regarding the role of "genomic medicine" in primary care. While many primary care physicians already incorporate genetic screening into their routine services, it has been predicted that new genetic tests and genetic treatments stemming from the Human Genome Project will become a routine component of primary care. The proposed study intends to investigate primary care physicians' perceptions of genomic medicine that may influence the adoption of this innovation into their primary care practice.

I am being invited to participate in this survey (along with 50 peers) because my name was recommended by an acquaintance.

My participation in this study entails answering this survey and returning it to Sandra Suther, in the enclosed self-addressed stamped envelope by _____. My participation is voluntary and I am not obligated to answer any of the questions posed in the questionnaire. As there is little information on this subject, however, my input will represent a valuable contribution to the study of primary care professionals' views of genomic medicine.

Some of the risks, discomforts and inconveniences that are reasonable to expect in this study are feeling that I haven't thought through some of the questions enough to provide answers, or feeling as if I don't have the "right" answer. On the other hand, some of the benefits that may reasonably be expected from participating in this study are: the opportunity to state my point-of-view on the subject and to reflect on my role as a health professional.

I understand that every effort will be made to keep all information confidential. My decision whether to participate will not affect my future relations with Texas A&M University, in any way. I understand I am under no obligation to participate in the study. I may withdraw from the study at any time, if I wish to.

This research study has been reviewed and approved by the Institutional Review Board – Human Subjects in Research, Texas A&M University. If I have any research-related problems or questions regarding subjects' rights I may contact the
Texas A & M Institutional Review Board through Dr. Michael W. Buckley, IRB Coordinator, Office of Vice President for Research at (979) 845-8585, mwbuckley@tamu.edu.

If, at any time, I have any questions about this study or further information I would like to add, I may contact Sandra Suther at the phone number or e-mail address below, or her advisor, Dr. Patricia Goodson at (979) 845-1756 or pgoodson@hlkn.tamu.edu.

I also understand that by filling out the survey and returning it by mail, I'm agreeing to participate in this study, and I may keep this copy of the informed consent.

PRINCIPAL INVESTIGATOR:_____
Sandra Suther, M.A.
Department of Health and Kinesiology
Texas A&M University
4243 TAMU
College Station TX 77843-4243
Phone: 979-696-0618
E-mail: ssuther@neo.tamu.edu

_____
SURVEY PARTICIPANT

_____
DATE

TEXAS A&M UNIVERSITY
College of Education and Human Development
Department of Health & Kinesiology

[Date]

Dear Dr.

I need your help with my dissertation research!

If you would take a few minutes to fill out this **short but very important** survey, it would be greatly appreciated. Results of the survey will be used to examine the factors that influence the likelihood of physicians incorporating genomic medicine into their primary care practice. This survey does **not** intend to measure knowledge about genetic medicine.

Your name was randomly selected from the Texas State Board of Medical Practitioners. All surveys will be number coded but your answers are completely confidential. Only statistical information from aggregated data will be used for reporting.

If you have any questions about the survey, feel free to call me or write me at the address below. If you prefer, the completed survey can be faxed back.

Thank you very much for helping with this important study.

Sincerely,

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TEXAS A&M UNIVERSITY
College of Education and Human Development
Department of Health & Kinesiology

Informed Consent for Survey Participants
Genomic Medicine in Primary Care: Texas Physicians' Adoption of an Innovation

April, 2003

I have been invited to participate in a study of the views that primary care physicians in Texas have of genomic medicine. Sandra Suther is conducting this study. Presently, she is a doctoral student in health education and health promotion in the Department of Health and Kinesiology, College of Education and Human Development, at Texas A&M University. One of her main research interests is public health genetics. This study is part of her dissertation research.

The recent completion of the first draft of the human genome sequence is generating concerns among health communities world-wide regarding the role of "genomic medicine" in primary care. While many primary care physicians already incorporate genetic screening into their routine services, it has been predicted that new genetic tests and genetic treatments stemming from the Human Genome Project will become a routine component of primary care. The proposed study intends to investigate primary care physicians' perceptions of genomic medicine that may influence the adoption of this innovation into their primary care practice.

I am being invited to participate in this survey (along with 1,350 peers) because my name was randomly chosen from the Texas State Board of Medical Examiners' directory of practitioners.

My participation in this study entails answering this survey and returning it to Sandra Suther, in the enclosed self-addressed stamped envelope by _____. My participation is voluntary and I am not obligated to answer any of the questions posed in the questionnaire. As there is little information on this subject, however, my input will represent a valuable contribution to the study of primary care professionals' views of genomic medicine.

Some of the risks, discomforts and inconveniences that are reasonable to expect in this study are feeling that I haven't thought through some of the questions enough to provide answers, or feeling as if I don't have the "right" answer. On the other hand, some of the benefits that may reasonably be expected from participating in this study are: the opportunity to state my point-of-view on the subject and to reflect on my role as a health professional.

I understand that every effort will be made to keep all information confidential. Even though surveys received a number-code so demographic differences between respondents and non-respondents can be assessed, these codes will only be available to the principal investigator, and will be kept in a locked file in her office until surveys are returned. After that, the list will be destroyed.

My decision whether to participate will not affect my future relations with Texas A&M University, in any way. I understand I am under no obligation to participate in the study. I may withdraw from the study at any time, if I wish to.

This research study has been reviewed and approved by the Institutional Review Board – Human Subjects in Research, Texas A&M University. If I have any research-related problems or questions regarding subjects' rights I may contact the
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Sandra Suther, M.A.
Department of Health and Kinesiology
Texas A&M University
4243 TAMU
College Station TX 77843-4243
Phone: 979-696-0618
E-mail: ssuther@neo.tamu.edu

_____
SURVEY PARTICIPANT

_____
DATE

VITA

Sandra Gayle Suther was born in Houston, Texas, on November 7, 1950, the daughter of Beatrice and Ausber E. Reynolds. She and her husband George have two children, Natalie and Christopher. After receiving a Bachelor of Arts degree in anthropology from The University of Texas at San Antonio in 1996, she enrolled in the Master of Arts program in anthropology to pursue her interest in medical anthropology. Employed as an ophthalmic technician during those years, she assisted in numerous medical/surgical missions in Mexico, which inspired her interest in culture and medicine. She also spent three years doing field research in the Rio Grande Valley of Texas and Mexico for a SPRANS funded Maternal and Child Health Genetics Project. Upon graduation in 2000 she applied and was accepted into the Health Education graduate program at Texas A&M University. While pursuing her doctoral degree, she had the opportunity to be involved with two research projects; a program evaluation of abstinence-only education programs in Texas, funded by the Texas Department of Health and a qualitative study of barriers to the provision of genetic services in primary care, funded by a grant from Texas A&M University. She was awarded outstanding graduate student 2002-2003.

Permanent Address: 12922 Reveille, San Antonio, Texas 78233